BEST AVAILABLE IMAGES

Defective images within this document are accurate representations of the original documents submitted by the applicant.

Defects in the images may include (but are not limited to):

- BLACK BORDERS
- TEXT CUT OFF AT TOP, BOTTOM OR SIDES
- FADED TEXT
- ILLEGIBLE TEXT
- SKEWED/SLANTED IMAGES
- COLORED PHOTOS
- BLACK OR VERY BLACK AND WHITE DARK PHOTOS
- GRAY SCALE DOCUMENTS

IMAGES ARE BEST AVAILABLE COPY.

As rescanning documents *will not* correct images, please do not report the images to the Image Problem Mailbox.

PUBMED 2594783
REMARK SEQUENCE FROM N.A., AND VARIANT CAIS MET-866.
REFERENCE 3 (residues 1 to 919)
AUTHORS Govindan,M.V.
TITLE Specific region in hormone binding domain is essential for hormone binding and trans-activation by human androgen receptor
JOURNAL Mol. Endocrinol. 4 (3), 417-427 (1990)
MEDLINE 90258935
PUBMED 2342476
REMARK SEQUENCE FROM N.A.
REFERENCE 4 (residues 1 to 919)
AUTHORS Chang,C.S., Kokontis,J. and Liao,S.T.
TITLE Structural analysis of complementary DNA and amino acid sequences of human and rat androgen receptors
JOURNAL Proc. Natl. Acad. Sci. U.S.A. 85 (19), 7211-7215 (1988)
MEDLINE 89017168
PUBMED 3174628
REMARK SEQUENCE FROM N.A.
TISSUE=Prostate
REFERENCE 5 (residues 1 to 919)
AUTHORS Tilley,W.D., Marcelli,M., Wilson,J.D. and McPhaul,M.J.
TITLE Characterization and expression of a cDNA encoding the human androgen receptor
JOURNAL Proc. Natl. Acad. Sci. U.S.A. 86 (1), 327-331 (1989)
MEDLINE 89098909
PUBMED 2911578
REMARK SEQUENCE FROM N.A.
TISSUE=Prostate
REFERENCE 6 (residues 1 to 919)
AUTHORS Marcelli,M., Tilley,W.D., Wilson,C.M., Griffin,J.E., Wilson,J.D. and McPhaul,M.J.
TITLE Definition of the human androgen receptor gene structure permits the identification of mutations that cause androgen resistance: premature termination of the receptor protein at amino acid residue 588 causes complete androgen resistance
JOURNAL Mol. Endocrinol. 4 (8), 1105-1116 (1990)
MEDLINE 91155943
PUBMED 2293020
REMARK SEQUENCE FROM N.A.
TISSUE=Prostate
REFERENCE 7 (residues 1 to 919)
AUTHORS Chang,C.S., Kokontis,J. and Liao,S.T.
TITLE Molecular cloning of human and rat complementary DNA encoding androgen receptors
JOURNAL Science 240 (4850), 324-326 (1988)
MEDLINE 88178111
PUBMED 3353726
REMARK SEQUENCE OF 189-919 FROM N.A.
REFERENCE 8 (residues 1 to 919)
AUTHORS Trapman,J., Klaassen,P., Kuiper,G.G., van der Korput,J.A., Faber,P.W., van Rooij,H.C., Geurts van Kessel,A., Voorhorst,M.M., Mulder,E. and Brinkmann,A.O.
TITLE Cloning, structure and expression of a cDNA encoding the human androgen receptor
JOURNAL Biochem. Biophys. Res. Commun. 153 (1), 241-248 (1988)
MEDLINE 88240407
PUBMED 3377788
REMARK SEQUENCE OF 468-919 FROM N.A.
REFERENCE 9 (residues 1 to 919)
AUTHORS Hsiao,P.W., Lin,D.L., Nakao,R. and Chang,C.

TITLE The linkage of Kennedy's neuron disease to ARA24, the first identified androgen receptor polyglutamine region-associated coactivator
JOURNAL J. Biol. Chem. 274 (29), 20229-20234 (1999)
MEDLINE 99329028
PUBMED 10400640
REMARK INTERACTION WITH RAN.
REFERENCE 10 (residues 1 to 919)
AUTHORS Sleddens,H.F.B.M., Oostra,B.A., Brinkmann,A.O. and Trapman,J.
TITLE Trinucleotide repeat polymorphism in the androgen receptor gene (AR)
JOURNAL Nucleic Acids Res. 20, 1427-1427 (1992)
REMARK POLYMORPHISM OF POLY-GLN REGION.
REFERENCE 11 (residues 1 to 919)
AUTHORS Lu,J. and Danielsen,M.
TITLE Direct Submission
JOURNAL Submitted (~FEB-1995)
REMARK POLYMORPHISM OF POLY-GLY REGION.
TISSUE=Blood
REFERENCE 12 (residues 1 to 919)
AUTHORS Giovannucci,E., Stampfer,M.J., Krithivas,K., Brown,M., Dahl,D., Brufsky,A., Talcott,J., Hennekens,C.H. and Kantoff,P.W.
TITLE The CAG repeat within the androgen receptor gene and its relationship to prostate cancer
JOURNAL Proc. Natl. Acad. Sci. U.S.A. 94 (7), 3320-3323 (1997)
MEDLINE 97250535
PUBMED 9096391
REMARK POLYMORPHISM OF POLY-GLN REGION.
REFERENCE 13 (residues 1 to 919)
AUTHORS Giovannucci,E., Stampfer,M.J., Krithivas,K., Brown,M., Dahl,D., Brufsky,A., Talcott,J., Hennekens,C.H. and Kantoff,P.W.
JOURNAL Proc. Natl. Acad. Sci. U.S.A. 94, 8272-8272 (1997)
REMARK ERRATUM.
REFERENCE 14 (residues 1 to 919)
AUTHORS Pinsky,L., Trifiro,M.A., Kaufman,M., Beitel,L.K., Mhatre,A., Kazemi-Esfarjani,P., Sabbaghian,N., Lumbroso,R., Alvarado,C., Vasiliou,M. and Gottlieb,B.
TITLE Androgen resistance due to mutation of the androgen receptor
JOURNAL Clin Invest Med 15 (5), 456-472 (1992)
MEDLINE 93092459
PUBMED 1458719
REMARK REVIEW ON VARIANTS.
REFERENCE 15 (residues 1 to 919)
AUTHORS Brown,T.R., Scherer,P.A., Chang,Y.-T., Migeon,C.J., Ghirri,P., Murono,K. and Zhou,Z.
TITLE Molecular genetics of human androgen insensitivity
JOURNAL Eur. J. Pediatr. 152 Suppl. 2, S62-S69 (1993)
REMARK REVIEW ON VARIANTS AIS.
REFERENCE 16 (residues 1 to 919)
AUTHORS Sultan,C., Lumbroso,S., Poujol,N., Belon,C., Boudon,C. and Lobaccaro,J.M.
TITLE Mutations of androgen receptor gene in androgen insensitivity syndromes
JOURNAL J. Steroid Biochem. Mol. Biol. 46 (5), 519-530 (1993)
MEDLINE 94059770
PUBMED 8240973
REMARK REVIEW ON VARIANTS.
REFERENCE 17 (residues 1 to 919)
AUTHORS Patterson,M.N., Hughes,I.A., Gottlieb,B. and Pinsky,L.
TITLE The androgen receptor gene mutations database

JOURNAL Nucleic Acids Res. 22 (17), 3560-3562 (1994)
MEDLINE 95023089
PUBMED 7937057
REMARK REVIEW ON VARIANTS.
REFERENCE 18 (residues 1 to 919)
AUTHORS Brinkmann,A.O., Jenster,G., Ris-Stalpers,C., van der Korput,J.A., Bruggenwirth,H.T., Boehmer,A.L. and Trapman,J.
TITLE Androgen receptor mutations
JOURNAL J. Steroid Biochem. Mol. Biol. 53 (1-6), 443-448 (1995)
MEDLINE 95352489
PUBMED 7626493
REMARK REVIEW ON VARIANTS.
REFERENCE 19 (residues 1 to 919)
AUTHORS Gottlieb,B., Trifiro,M., Lumbroso,R. and Pinsky,L.
TITLE The androgen receptor gene mutations database
JOURNAL Nucleic Acids Res. 25 (1), 158-162 (1997)
MEDLINE 97169385
PUBMED 9016528
REMARK REVIEW ON VARIANTS.
REFERENCE 20 (residues 1 to 919)
AUTHORS Veldscholte,J., Ris-Stalpers,C., Kuiper,G.G., Jenster,G., Berrevoets,C., Claassen,E., van Rooij,H.C., Trapman,J., Brinkmann,A.O. and Mulder,E.
TITLE A mutation in the ligand binding domain of the androgen receptor of human LNCaP cells affects steroid binding characteristics and response to anti-androgens
JOURNAL Biochem. Biophys. Res. Commun. 173 (2), 534-540 (1990)
MEDLINE 91083633
PUBMED 2260966
REMARK VARIANT LNCAP ALA-877.
REFERENCE 21 (residues 1 to 919)
AUTHORS Brown,T.R., Lubahn,D.B., Wilson,E.M., French,F.S., Migeon,C.J. and Corden,J.L.
TITLE Functional characterization of naturally occurring mutant androgen receptors from subjects with complete androgen insensitivity
JOURNAL Mol. Endocrinol. 4 (12), 1759-1772 (1990)
MEDLINE 91186983
PUBMED 2082179
REMARK VARIANTS CAIS CYS-774; GLN-831 AND MET-866.
REFERENCE 22 (residues 1 to 919)
AUTHORS Marcelli,M., Tilley,W.D., Zoppi,S., Griffin,J.E., Wilson,J.D. and McPhaul,M.J.
TITLE Androgen resistance associated with a mutation of the androgen receptor at amino acid 772 (Arg----Cys) results from a combination of decreased messenger ribonucleic acid levels and impairment of receptor function
JOURNAL J. Clin. Endocrinol. Metab. 73 (2), 318-325 (1991)
MEDLINE 91310758
PUBMED 1856263
REMARK VARIANT CYS-774.
REFERENCE 23 (residues 1 to 919)
AUTHORS Marcelli,M., Zoppi,S., Grino,P.B., Griffin,J.E., Wilson,J.D. and McPhaul,M.J.
TITLE A mutation in the DNA-binding domain of the androgen receptor gene causes complete testicular feminization in a patient with receptor-positive androgen resistance
JOURNAL J. Clin. Invest. 87 (3), 1123-1126 (1991)
MEDLINE 91154385
PUBMED 1999491
REMARK VARIANT CAIS PRO-617.

REFERENCE 24 (residues 1 to 919)
AUTHORS McPhaul,M.J., Marcelli,M., Tilley,W.D., Griffin,J.E., Isidro-Gutierrez,R.F. and Wilson,J.D.
TITLE Molecular basis of androgen resistance in a family with a qualitative abnormality of the androgen receptor and responsive to high-dose androgen therapy
JOURNAL J. Clin. Invest. 87 (4), 1413-1421 (1991)
MEDLINE 91185626
PUBMED 2010552
REMARK VARIANT PAIS CYS-763.
REFERENCE 25 (residues 1 to 919)
AUTHORS Ris-Stalpers,C., Trifiro,M.A., Kuiper,G.G.J.M., Jenster,G., Romalo,G., Sai,T., van Rooij,H.C.J., Kaufman,M., Rosenfield,R.L., Liao,S., Schweikert,H.-U., Trapman,J., Pinsky,L. and Brinkmann,A.O.
TITLE Substitution of aspartic acid-686 by histidine or asparagine in the human androgen receptor leads to a functionally inactive protein with altered hormone-binding characteristics
JOURNAL Mol. Endocrinol. 5 (10), 1562-1569 (1991)
MEDLINE 92131007
PUBMED 1775137
REMARK VARIANTS CAIS ASN-695 AND HIS-695, AND SEQUENCE OF 629-723 FROM N.A.
REFERENCE 26 (residues 1 to 919)
AUTHORS La Spada,A.R., Wilson,E.M., Lubahn,D.B., Harding,A.E. and Fischbeck,K.H.
TITLE Androgen receptor gene mutations in X-linked spinal and bulbar muscular atrophy
JOURNAL Nature 352 (6330), 77-79 (1991)
MEDLINE 91287825
PUBMED 2062380
REMARK VARIANTS SBMA IN POLY-GLN REGION.
REFERENCE 27 (residues 1 to 919)
AUTHORS Prior,L., Bordet,S., Trifiro,M.A., Mhatre,A., Kaufman,M., Pinsky,L., Wrogemann,K., Belsham,D.D., Pereira,F., Greenberg,C.R., Trapman,J., Brinkmann,A.O., Chang,C. and Liao,S.
TITLE Replacement of arginine 773 by cysteine or histidine in the human androgen receptor causes complete androgen insensitivity with different receptor phenotypes
JOURNAL Am. J. Hum. Genet. 51 (1), 143-155 (1992)
MEDLINE 92303560
PUBMED 1609793
REMARK VARIANTS CAIS CYS-774 AND HIS-774.
REFERENCE 28 (residues 1 to 919)
AUTHORS Saunders,P.T., Padayachi,T., Tincello,D.G., Shalet,S.M. and Wu,F.C.
TITLE Point mutations detected in the androgen receptor gene of three men with partial androgen insensitivity syndrome
JOURNAL Clin. Endocrinol. (Oxf) 37 (3), 214-220 (1992)
MEDLINE 93047389
PUBMED 1424203
REMARK VARIANTS PAIS LYS-608 AND LEU-866.
REFERENCE 29 (residues 1 to 919)
AUTHORS Sweet,C.R., Behzadian,M.A. and McDonough,P.G.
TITLE A unique point mutation in the androgen receptor gene in a family with complete androgen insensitivity syndrome
JOURNAL Fertil. Steril. 58 (4), 703-707 (1992)
MEDLINE 93050279
PUBMED 1426313
REMARK VARIANT CAIS THR-765.
REFERENCE 30 (residues 1 to 919)
AUTHORS Jakubiczka,S., Werder,E.A. and Wieacker,P.

TITLE Point mutation in the steroid-binding domain of the androgen receptor gene in a family with complete androgen insensitivity syndrome (CAIS)
JOURNAL Hum. Genet. 90 (3), 311-312 (1992)
MEDLINE 93138625
PUBMED 1487249
REMARK VARIANT CAIS VAL-749.
REFERENCE 31 (residues 1 to 919)
AUTHORS Batch,J.A., Williams,D.M., Davies,H.R., Brown,B.D., Evans,B.A., Hughes,I.A. and Patterson,M.N.
TITLE Androgen receptor gene mutations identified by SSCP in fourteen subjects with androgen insensitivity syndrome
JOURNAL Hum. Mol. Genet. 1 (7), 497-503 (1992)
MEDLINE 93338440
PUBMED 1307250
REMARK VARIANTS CAIS, AND VARIANTS PAIS.
REFERENCE 32 (residues 1 to 919)
AUTHORS Nakao,R., Haji,M., Yanase,T., Ogo,A., Takayanagi,R., Katsube,T., Fukumaki,Y. and Nawata,H.
TITLE A single amino acid substitution (Met786----Val) in the steroid-binding domain of human androgen receptor leads to complete androgen insensitivity syndrome
JOURNAL J. Clin. Endocrinol. Metab. 74 (5), 1152-1157 (1992)
MEDLINE 92235226
PUBMED 1569163
REMARK VARIANT CAIS VAL-787.
REFERENCE 33 (residues 1 to 919)
AUTHORS Wilson,C.M., Griffin,J.E., Wilson,J.D., Marcelli,M., Zoppi,S. and McPhaul,M.J.
TITLE Immunoreactive androgen receptor expression in subjects with androgen resistance
JOURNAL J. Clin. Endocrinol. Metab. 75 (6), 1474-1478 (1992)
MEDLINE 93100381
PUBMED 1464650
REMARK VARIANTS CAIS ARG-741 AND CYS-834.
REFERENCE 34 (residues 1 to 919)
AUTHORS McPhaul,M.J., Marcelli,M., Zoppi,S., Wilson,C.M., Griffin,J.E. and Wilson,J.D.
TITLE Mutations in the ligand-binding domain of the androgen receptor gene cluster in two regions of the gene
JOURNAL J. Clin. Invest. 90 (5), 2097-2101 (1992)
MEDLINE 93055453
PUBMED 1430233
REMARK VARIANTS CAIS, AND VARIANTS PAIS.
REFERENCE 35 (residues 1 to 919)
AUTHORS Veldscholte,J., Berrevoets,C.A., Ris-Stalpers,C., Kuiper,G.G., Jenster,G., Trapman,J., Brinkmann,A.O. and Mulder,E.
TITLE The androgen receptor in LNCaP cells contains a mutation in the ligand binding domain which affects steroid binding characteristics and response to antiandrogens
JOURNAL J. Steroid Biochem. Mol. Biol. 41 (3-8), 665-669 (1992)
MEDLINE 92222955
PUBMED 1562539
REMARK VARIANT PROSTATE CANCER ALA-877.
REFERENCE 36 (residues 1 to 919)
AUTHORS Zoppi,S., Marcelli,M., Deslypere,J.P., Griffin,J.E., Wilson,J.D. and McPhaul,M.J.
TITLE Amino acid substitutions in the DNA-binding domain of the human androgen receptor are a frequent cause of receptor-binding positive androgen resistance

JOURNAL Mol. Endocrinol. 6 (3), 409-415 (1992)
MEDLINE 92261595
PUBMED 1316540
REMARK VARIANTS CAIS TYR-559 AND ARG-576, AND VARIANTS PAIS GLY-597 AND PRO-617.
REFERENCE 37 (residues 1 to 919)
AUTHORS De Bellis,A., Quigley,C.A., Cariello,N.F., el-Awady,M.K., Sar,M., Lane,M.V., Wilson,E.M. and French,F.S.
TITLE Single base mutations in the human androgen receptor gene causing complete androgen insensitivity: rapid detection by a modified denaturing gradient gel electrophoresis technique
JOURNAL Mol. Endocrinol. 6 (11), 1909-1920 (1992)
MEDLINE 93125565
PUBMED 1480178
REMARK VARIANTS CAIS SER-705; VAL-749; PHE-759; HIS-774; CYS-855 AND GLY-864.
REFERENCE 38 (residues 1 to 919)
AUTHORS Wooster,R., Mangion,J., Eeles,R., Smith,S., Dowsett,M., Averill,D., Barrett-Lee,P., Easton,D.F., Ponder,B.A. and Stratton,M.R.
TITLE A germline mutation in the androgen receptor gene in two brothers with breast cancer and Reifenstein syndrome
JOURNAL Nat. Genet. 2 (2), 132-134 (1992)
MEDLINE 93265137
PUBMED 1303262
REMARK VARIANT PAIS/BREAST CANCER GLN-607.
REFERENCE 39 (residues 1 to 919)
AUTHORS Newmark,J.R., Hardy,D.O., Tonb,D.C., Carter,B.S., Epstein,J.I., Isaacs,W.B., Brown,T.R. and Barrack,E.R.
TITLE Androgen receptor gene mutations in human prostate cancer
JOURNAL Proc. Natl. Acad. Sci. U.S.A. 89 (14), 6319-6323 (1992)
MEDLINE 92335289
PUBMED 1631125
REMARK VARIANT MET-730.
REFERENCE 40 (residues 1 to 919)
AUTHORS Macke,J.P., Hu,N., Hu,S., Bailey,M., King,V.L., Brown,T., Hamer,D. and Nathans,J.
TITLE Sequence variation in the androgen receptor gene is not a common determinant of male sexual orientation
JOURNAL Am. J. Hum. Genet. 53 (4), 844-852 (1993)
MEDLINE 94027050
PUBMED 8213813
REMARK VARIANTS ARG-205 AND ASP-793.
REFERENCE 41 (residues 1 to 919)
AUTHORS Lumbroso,S., Lobaccaro,J.M., Belon,C., Martin,D., Chaussain,J.L. and Sultan,C.
TITLE A new mutation within the deoxyribonucleic acid-binding domain of the androgen receptor gene in a family with complete androgen insensitivity syndrome
JOURNAL Fertil. Steril. 60 (5), 814-819 (1993)
MEDLINE 94039857
PUBMED 8224266
REMARK VARIANT CAIS PHE-581.
REFERENCE 42 (residues 1 to 919)
AUTHORS Lobaccaro,J.M., Lumbroso,S., Ktari,R., Dumas,R. and Sultan,C.
TITLE An exonic point mutation creates a MaeIII site in the androgen receptor gene of a family with complete androgen insensitivity syndrome
JOURNAL Hum. Mol. Genet. 2 (7), 1041-1043 (1993)
MEDLINE 93372806
PUBMED 8103398

REMARK VARIANT CAIS VAL-754.
REFERENCE 43 (residues 1 to 919)
AUTHORS Lobaccaro,J.M., Lumbroso,S., Belon,C., Galtier-Dereure,F., Bringer,J., Lesimple,T., Namer,M., Cutuli,B.F., Pujol,H. and Sultan,C.
TITLE Androgen receptor gene mutation in male breast cancer
JOURNAL Hum. Mol. Genet. 2 (11), 1799-1802 (1993)
MEDLINE 94108428
PUBMED 8281139
REMARK VARIANT PAIS/BREAST CANCER LYS-608.
REFERENCE 44 (residues 1 to 919)
AUTHORS Adeyemo,O., Kallio,P.J., Palvimo,J.J., Kontula,K. and Janne,O.A.
TITLE A single-base substitution in exon 6 of the androgen receptor gene causing complete androgen insensitivity: the mutated receptor fails to transactivate but binds to DNA in vitro
JOURNAL Hum. Mol. Genet. 2 (11), 1809-1812 (1993)
MEDLINE 94108430
PUBMED 8281140
REMARK VARIANT CAIS ARG-807.
REFERENCE 45 (residues 1 to 919)
AUTHORS Nakao,R., Yanase,T., Sakai,Y., Haji,M. and Nawata,H.
TITLE A single amino acid substitution (gly743 --> val) in the steroid-binding domain of the human androgen receptor leads to Reifenstein syndrome
JOURNAL J. Clin. Endocrinol. Metab. 77 (1), 103-107 (1993)
MEDLINE 93315568
PUBMED 8325932
REMARK VARIANT PAIS VAL-743.
REFERENCE 46 (residues 1 to 919)
AUTHORS Hiort,O., Huang,Q., Sinnecker,G.H., Sadeghi-Nejad,A., Kruse,K., Wolfe,H.J. and Yandell,D.W.
TITLE Single strand conformation polymorphism analysis of androgen receptor gene mutations in patients with androgen insensitivity syndromes: application for diagnosis, genetic counseling, and therapy
JOURNAL J. Clin. Endocrinol. Metab. 77 (1), 262-266 (1993)
MEDLINE 93315600
PUBMED 8325950
REMARK VARIANTS CAIS LYS-681 AND THR-842, AND VARIANTS PAIS HIS-840 AND LEU-866.
REFERENCE 47 (residues 1 to 919)
AUTHORS Batch,J.A., Evans,B.A., Hughes,I.A. and Patterson,M.N.
TITLE Mutations of the androgen receptor gene identified in perineal hypospadias
JOURNAL J. Med. Genet. 30 (3), 198-201 (1993)
MEDLINE 93233131
PUBMED 8097257
REMARK VARIANTS PAIS HIS-855 AND MET-869.
REFERENCE 48 (residues 1 to 919)
AUTHORS Lobaccaro,J.M., Lumbroso,S., Berta,P., Chaussain,J.L. and Sultan,C.
TITLE Complete androgen insensitivity syndrome associated with a de novo mutation of the androgen receptor gene detected by single strand conformation polymorphism
JOURNAL J. Steroid Biochem. Mol. Biol. 44 (3), 211-216 (1993)
MEDLINE 93213715
PUBMED 8096390
REMARK VARIANT CAIS VAL-743.
REFERENCE 49 (residues 1 to 919)
AUTHORS Suzuki,H., Sato,N., Watabe,Y., Masai,M., Seino,S. and Shimazaki,J.
TITLE Androgen receptor gene mutations in human prostate cancer

JOURNAL J. Steroid Biochem. Mol. Biol. 46 (6), 759-765 (1993)
MEDLINE 94100129
PUBMED 8274409
REMARK VARIANTS PROSTATE CANCER HIS-701 AND ALA-877.
REFERENCE 50 (residues 1 to 919)
AUTHORS Kazemi-Esfarjani,P., Beitel,L.K., Trifiro,M., Kaufman,M., Rennie,P., Sheppard,P., Matusik,R. and Pinsky,L.
TITLE Substitution of valine-865 by methionine or leucine in the human androgen receptor causes complete or partial androgen insensitivity, respectively with distinct androgen receptor phenotypes
JOURNAL Mol. Endocrinol. 7 (1), 37-46 (1993)
MEDLINE 93188862
PUBMED 8446106
REMARK VARIANT CAIS MET-866, AND VARIANT PAIS LEU-866.
REFERENCE 51 (residues 1 to 919)
AUTHORS Mowszowicz,I., Lee,H.J., Chen,H.T., Mestayer,C., Portois,M.C., Cabrol,S., Mauvais-Jarvis,P. and Chang,C.
TITLE A point mutation in the second zinc finger of the DNA-binding domain of the androgen receptor gene causes complete androgen insensitivity in two siblings with receptor-positive androgen resistance
JOURNAL Mol. Endocrinol. 7 (7), 861-869 (1993)
MEDLINE 94019395
PUBMED 8413310
REMARK VARIANT CAIS HIS-615.
REFERENCE 52 (residues 1 to 919)
AUTHORS Culig,Z., Hobisch,A., Cronauer,M.V., Cato,A.C., Hittmair,A., Radmayr,C., Eberle,J., Bartsch,G. and Klocker,H.
TITLE Mutant androgen receptor detected in an advanced-stage prostatic carcinoma is activated by adrenal androgens and progesterone
JOURNAL Mol. Endocrinol. 7 (12), 1541-1550 (1993)
MEDLINE 94195311
PUBMED 8145761
REMARK VARIANT PROSTATE CANCER MET-715.
REFERENCE 53 (residues 1 to 919)
AUTHORS Lobaccaro,J.-M., Lumbroso,S., Belon,C., Chaussain,J.L., Toublanc,J.E., Leheup,B. and Sultan,C.
TITLE Androgen receptor (AR) gene mutations in 6 families with androgen insensitivity syndrome (Abstract #114)
JOURNAL Pediatr. Res. Suppl. 33, S22-S22 (1993)
REMARK VARIANTS CAIS PHE-581; VAL-743; VAL-754; GLU-767 AND CYS-855.
REFERENCE 54 (residues 1 to 919)
AUTHORS Castagnaro,M., Yandell,D.W., Dockhorn-Dworniczak,B., Wolfe,H.J. and Poremba,C.
TITLE Androgen receptor gene mutations and p53 gene analysis in advanced prostate cancer
JOURNAL Verh Dtsch Ges Pathol 77, 119-123 (1993)
MEDLINE 94189145
PUBMED 7511268
REMARK VARIANTS PROSTATE CANCER LEU-340 AND GLU-798.
REFERENCE 55 (residues 1 to 919)
AUTHORS Schoenberg,M.P., Hakimi,J.M., Wang,S., Bova,G.S., Epstein,J.I., Fischbeck,K.H., Isaacs,W.B., Walsh,P.C. and Barrack,E.R.
TITLE Microsatellite mutation (CAG24-->18) in the androgen receptor gene in human prostate cancer
JOURNAL Biochem. Biophys. Res. Commun. 198 (1), 74-80 (1994)
MEDLINE 94121667
PUBMED 8292051
REMARK VARIANT PROSTATE CANCER IN POLY-GLN REGION.

REFERENCE 56 (residues 1 to 919)
AUTHORS Gaddipati,J.P., McLeod,D.G., Heidenberg,H.B., Sesterhenn,I.A., Finger,M.J., Moul,J.W. and Srivastava,S.
TITLE Frequent detection of codon 877 mutation in the androgen receptor gene in advanced prostate cancers
JOURNAL Cancer Res. 54 (11), 2861-2864 (1994)
MEDLINE 94243798
PUBMED 8187068
REMARK VARIANT PROSTATE CANCER ALA-877.
REFERENCE 57 (residues 1 to 919)
AUTHORS Lobaccaro,J.M., Belon,C., Lumbroso,S., Olewniczack,G., Carre-Pigeon,F., Job,J.C., Chaussain,J.L., Toublanc,J.E. and Sultan,C.
TITLE Molecular prenatal diagnosis of partial androgen insensitivity syndrome based on the Hind III polymorphism of the androgen receptor gene
JOURNAL Clin. Endocrinol. (Oxf) 40 (3), 297-302 (1994)
MEDLINE 94244054
PUBMED 7910529
REMARK VARIANT PAIS TRP-568.
REFERENCE 58 (residues 1 to 919)
AUTHORS Lumbroso,S., Lobaccaro,J.M., Belon,C., Amram,S., Bachelard,B., Garandeau,P. and Sultan,C.
TITLE Molecular prenatal exclusion of familial partial androgen insensitivity (Reifenstein syndrome)
JOURNAL Eur. J. Endocrinol. 130 (4), 327-332 (1994)
MEDLINE 94214640
PUBMED 7909256
REMARK VARIANT PAIS HIS-840.
REFERENCE 59 (residues 1 to 919)
AUTHORS Imasaki,K., Hasegawa,T., Okabe,T., Sakai,Y., Haji,M., Takayanagi,R. and Nawata,H.
TITLE Single amino acid substitution (840Arg-->His) in the hormone-binding domain of the androgen receptor leads to incomplete androgen insensitivity syndrome associated with a thermolabile androgen receptor
JOURNAL Eur. J. Endocrinol. 130 (6), 569-574 (1994)
MEDLINE 94264834
PUBMED 8205256
REMARK VARIANT PAIS HIS-840.
REFERENCE 60 (residues 1 to 919)
AUTHORS Hiort,O., Klauber,G., Cendron,M., Sinnecker,G.H., Keim,L., Schwinger,E., Wolfe,H.J. and Yandell,D.W.
TITLE Molecular characterization of the androgen receptor gene in boys with hypospadias
JOURNAL Eur. J. Pediatr. 153 (5), 317-321 (1994)
MEDLINE 94307287
PUBMED 8033918
REMARK VARIANT PAIS VAL-870.
REFERENCE 61 (residues 1 to 919)
AUTHORS Schwartz,M., Skovby,F., Mueller,J., Nielsen,O. and Skakkebaek,N.E.
TITLE Partial androgen insensitivity (PAIS) in a large eskimo kindred caused by a delD690 mutation in the androgen receptor (AR) gene (Abstract #244)
JOURNAL Horm. Res. 41, 117-117 (1994)
REMARK VARIANT PAIS ASP-690 DEL.
REFERENCE 62 (residues 1 to 919)
AUTHORS Beitel,L.K., Prior,L., Vasiliou,D.M., Gottlieb,B., Kaufman,M., Lumbroso,R., Alvarado,C., McGillivray,B., Trifiro,M. and Pinsky,L.
TITLE Complete androgen insensitivity due to mutations in the probable

alpha-helical segments of the DNA-binding domain in the human androgen receptor
JOURNAL Hum. Mol. Genet. 3 (1), 21-27 (1994)
MEDLINE 94214451
PUBMED 8162033
REMARK VARIANTS CAIS PHE-582 DEL; ARG-615 DEL AND HIS-615.
REFERENCE 63 (residues 1 to 919)
AUTHORS Hiort,O., Wodtke,A., Struve,D., Zollner,A. and Sinnecker,G.H.
TITLE Detection of point mutations in the androgen receptor gene using non-isotopic single strand conformation polymorphism analysis. German Collaborative Intersex Study Group
JOURNAL Hum. Mol. Genet. 3 (7), 1163-1166 (1994)
MEDLINE 95072584
PUBMED 7981687
REMARK VARIANTS PAIS SER-582; TYR-604; ALA-708; LEU-754 AND HIS-771, AND VARIANT CAIS TRP-779.
REFERENCE 64 (residues 1 to 919)
AUTHORS Baldazzi,L., Baroncini,C., Pirazzoli,P., Balsamo,A., Capelli,M., Marchetti,G., Bernardi,F. and Cacciari,E.
TITLE Two mutations causing complete androgen insensitivity: a frame-shift in the steroid binding domain and a Cys-->Phe substitution in the second zinc finger of the androgen receptor
JOURNAL Hum. Mol. Genet. 3 (7), 1169-1170 (1994)
MEDLINE 95072586
PUBMED 7981689
REMARK VARIANT CAIS PHE-601.
REFERENCE 65 (residues 1 to 919)
AUTHORS De Bellis,A., Quigley,C.A., Marschke,K.B., el-Awady,M.K., Lane,M.V., Smith,E.P., Sar,M., Wilson,E.M. and French,F.S.
TITLE Characterization of mutant androgen receptors causing partial androgen insensitivity syndrome
JOURNAL J. Clin. Endocrinol. Metab. 78 (3), 513-522 (1994)
MEDLINE 94171952
PUBMED 8126121
REMARK VARIANTS PAIS ARG-616; HIS-840 AND MET-889.
REFERENCE 66 (residues 1 to 919)
AUTHORS Tsukada,T., Inoue,M., Tachibana,S., Nakai,Y. and Takebe,H.
TITLE An androgen receptor mutation causing androgen resistance in undervirilized male syndrome
JOURNAL J. Clin. Endocrinol. Metab. 79 (4), 1202-1207 (1994)
MEDLINE 95051264
PUBMED 7962294
REMARK VARIANT MAIS PHE-790.
REFERENCE 67 (residues 1 to 919)
AUTHORS Beitel,L.K., Kazemi-Esfarjani,P., Kaufman,M., Lumbroso,R., DiGeorge,A.M., Killinger,D.W., Trifiro,M.A. and Pinsky,L.
TITLE Substitution of arginine-839 by cysteine or histidine in the androgen receptor causes different receptor phenotypes in cultured cells and coordinate degrees of clinical androgen resistance
JOURNAL J. Clin. Invest. 94 (2), 546-554 (1994)
MEDLINE 94314956
PUBMED 8040309
REMARK VARIANTS PAIS CYS-840 AND HIS-840.
REFERENCE 68 (residues 1 to 919)
AUTHORS Marcelli,M., Zoppi,S., Wilson,C.M., Griffin,J.E. and McPhaul,M.J.
TITLE Amino acid substitutions in the hormone-binding domain of the human androgen receptor alter the stability of the hormone receptor complex
JOURNAL J. Clin. Invest. 94 (4), 1642-1650 (1994)
MEDLINE 95015035

PUBMED 7929841
REMARK VARIANTS CAIS, AND VARIANTS PAIS.
REFERENCE 69 (residues 1 to 919)
AUTHORS Yong,E.L., Ng,S.C., Roy,A.C., Yun,G. and Ratnam,S.S.
TITLE Pregnancy after hormonal correction of severe spermatogenic defect due to mutation in androgen receptor gene
JOURNAL Lancet 344 (8925), 826-827 (1994)
MEDLINE 94366236
PUBMED 7993455
REMARK VARIANT MAIS LYS-727.
REFERENCE 70 (residues 1 to 919)
AUTHORS Ris-Stalpers,C., Hoogenboezem,T., Sleddens,H.F.B.M., Verleun-Mooijman,M.C.T., Degenhart,H.J., Drop,S.L.S., Halley,D.J.J., Oosterwijk,J.C., Hodgins,M.B., Trapman,J. and Brinkmann,A.O.
TITLE A practical approach to the detection of androgen receptor gene mutations and pedigree analysis in families with x-linked androgen insensitivity
JOURNAL Pediatr. Res. 36 (2), 227-234 (1994)
MEDLINE 95061051
PUBMED 7970939
REMARK VARIANTS CAIS HIS-615 AND LEU-764, AND VARIANTS PAIS VAL-742 AND THR-745.
REFERENCE 71 (residues 1 to 919)
AUTHORS Imai,A., Ohno,T., Iida,K., Ohsuye,K., Okano,Y. and Tamaya,T.
TITLE A frame-shift mutation of the androgen receptor gene in a patient with receptor-negative complete testicular feminization: comparison with a single base substitution in a receptor-reduced incomplete form
JOURNAL Ann Clin Biochem 32 (Pt 5), 482-486 (1995)
MEDLINE 96113133
PUBMED 8830623
REMARK VARIANT PAIS HIS-840.
REFERENCE 72 (residues 1 to 919)
AUTHORS Takahashi,H., Furusato,M., Allsbrook,W.C. Jr., Nishii,H., Wakui,S., Barrett,J.C. and Boyd,J.
TITLE Prevalence of androgen receptor gene mutations in latent prostatic carcinomas from Japanese men
JOURNAL Cancer Res. 55 (8), 1621-1624 (1995)
MEDLINE 95228039
PUBMED 7712463
REMARK VARIANTS PROSTATE CANCER.
REFERENCE 73 (residues 1 to 919)
AUTHORS Davies,H.R., Hughes,I.A. and Patterson,M.N.
TITLE Genetic counselling in complete androgen insensitivity syndrome: trinucleotide repeat polymorphisms, single-strand conformation polymorphism and direct detection of two novel mutations in the androgen receptor gene
JOURNAL Clin. Endocrinol. (Oxf) 43 (1), 69-77 (1995)
MEDLINE 95368867
PUBMED 7641413
REMARK VARIANT CAIS VAL-881.
REFERENCE 74 (residues 1 to 919)
AUTHORS Quigley,C.A., De Bellis,A., Marschke,K.B., el-Awady,M.K., Wilson,E.M. and French,F.S.
TITLE Androgen receptor defects: historical, clinical, and molecular perspectives
JOURNAL Endocr. Rev. 16 (3), 271-321 (1995)
MEDLINE 95401910
PUBMED 7671849

REMARK VARIANTS CAIS SER-705 AND HIS-763, AND VARIANTS PAIS LEU-725; THR-737; HIS-774 AND GLU-798.
REFERENCE 75 (residues 1 to 919)
AUTHORS Quigley,C.A., De Bellis,A., Marschke,K.B., el-Awady,M.K., Wilson,E.M. and French,F.S.
JOURNAL Endocr. Rev. 1995 16, 546-546 (1995)
REMARK ERRATUM.
REFERENCE 76 (residues 1 to 919)
AUTHORS Shkolny,D.L., Brown,T.R., Punnett,H.H., Kaufman,M., Trifiro,M.A. and Pinsky,L.
TITLE Characterization of alternative amino acid substitutions at arginine 830 of the androgen receptor that cause complete androgen insensitivity in three families
JOURNAL Hum. Mol. Genet. 4 (4), 515-521 (1995)
MEDLINE 95359952
PUBMED 7633398
REMARK VARIANTS CAIS LEU-831 AND GLN-831.
REFERENCE 77 (residues 1 to 919)
AUTHORS Belsham,D.D., Pereira,F., Greenberg,C.R., Liao,S. and Wrogemann,K.
TITLE Leu-676-Pro mutation of the androgen receptor causes complete androgen insensitivity syndrome in a large Hutterite kindred
JOURNAL Hum. Mutat. 5 (1), 28-33 (1995)
MEDLINE 95245337
PUBMED 7537149
REMARK VARIANT CAIS PRO-677.
REFERENCE 78 (residues 1 to 919)
AUTHORS Murono,K., Mendonca,B.B., Arnhold,I.J., Rigon,A.C., Migeon,C.J. and Brown,T.R.
TITLE Human androgen insensitivity due to point mutations encoding amino acid substitutions in the androgen receptor steroid-binding domain
JOURNAL Hum. Mutat. 6 (2), 152-162 (1995)
MEDLINE 96047148
PUBMED 7581399
REMARK VARIANT PAIS CYS-763, AND VARIANTS CAIS TRP-779; VAL-807 AND CYS-855.
REFERENCE 79 (residues 1 to 919)
AUTHORS Peterziel,H., Culig,Z., Stober,J., Hobisch,A., Radmayr,C., Bartsch,G., Klocker,H. and Cato,A.C.
TITLE Mutant androgen receptors in prostatic tumors distinguish between amino-acid-sequence requirements for transactivation and ligand binding
JOURNAL Int. J. Cancer 63 (4), 544-550 (1995)
MEDLINE 96074743
PUBMED 7591265
REMARK VARIANT PROSTATE CANCER MET-730.
REFERENCE 80 (residues 1 to 919)
AUTHORS Allera,A., Herbst,M.A., Griffin,J.E., Wilson,J.D., Schweikert,H.U. and McPhaul,M.J.
TITLE Mutations of the androgen receptor coding sequence are infrequent in patients with isolated hypospadias
JOURNAL J. Clin. Endocrinol. Metab. 80 (9), 2697-2699 (1995)
MEDLINE 95403657
PUBMED 7673412
REMARK VARIANT PAIS VAL-568.
REFERENCE 81 (residues 1 to 919)
AUTHORS Elo,J.P., Kvist,L., Leinonen,K., Isomaa,V., Henttu,P., Lukkarinen,O. and Vihko,P.
TITLE Mutated human androgen receptor gene detected in a prostatic cancer patient is also activated by estradiol
JOURNAL J. Clin. Endocrinol. Metab. 80 (12), 3494-3500 (1995)

MEDLINE 96094386
PUBMED 8530589
REMARK VARIANT PROSTATE CANCER LEU-726.
REFERENCE 82 (residues 1 to 919)
AUTHORS Gast,A., Neuschmid-Kaspar,F., Klocker,H. and Cato,A.C.
TITLE A single amino acid exchange abolishes dimerization of the androgen receptor and causes Reifenstein syndrome
JOURNAL Mol. Cell. Endocrinol. 111 (1), 93-98 (1995)
MEDLINE 95377517
PUBMED 7649358
REMARK VARIANT PAIS THR-596.
REFERENCE 83 (residues 1 to 919)
AUTHORS Taplin,M.E., Bubley,G.J., Shuster,T.D., Frantz,M.E., Spooner,A.E., Ogata,G.K., Keer,H.N. and Balk,S.P.
TITLE Mutation of the androgen-receptor gene in metastatic androgen-independent prostate cancer
JOURNAL N. Engl. J. Med. 332 (21), 1393-1398 (1995)
MEDLINE 95240676
PUBMED 7723794
REMARK VARIANTS PROSTATE CANCER.
REFERENCE 84 (residues 1 to 919)
AUTHORS Hiort,O., Sinnecker,G.H., Holterhus,P.M., Nitsche,E.M. and Kruse,K.
TITLE The clinical and molecular spectrum of androgen insensitivity syndromes
JOURNAL Am. J. Med. Genet. 63 (1), 218-222 (1996)
MEDLINE 96298278
PUBMED 8723113
REMARK VARIANTS CAIS AND PAIS.
REFERENCE 85 (residues 1 to 919)
AUTHORS Tilley,W.D., Buchanan,G., Hickey,T.E. and Bentel,J.M.
TITLE Mutations in the androgen receptor gene are associated with progression of human prostate cancer to androgen independence
JOURNAL Clin. Cancer Res. 2 (2), 277-285 (1996)
MEDLINE 99035018
PUBMED 9816170
REMARK VARIANTS PROSTATE CANCER.
REFERENCE 86 (residues 1 to 919)
AUTHORS Weidemann,W., Linck,B., Haupt,H., Mentrup,B., Romalo,G., Stockklauser,K., Brinkmann,A.O., Schweikert,H.U. and Spindler,K.D.
TITLE Clinical and biochemical investigations and molecular analysis of subjects with mutations in the androgen receptor gene
JOURNAL Clin. Endocrinol. (Oxf) 45 (6), 733-739 (1996)
MEDLINE 97191382
PUBMED 9039340
REMARK VARIANTS PAIS GLN-607; THR-610; LEU-754; HIS-840; THR-842 AND HIS-855, AND VARIANT CAIS MET-866.
REFERENCE 87 (residues 1 to 919)
AUTHORS Malmgren,H., Gustavsson,J., Tuvemo,T. and Dahl,N.
TITLE Rapid detection of a mutation hot-spot in the human androgen receptor
JOURNAL Clin. Genet. 50 (4), 202-205 (1996)
MEDLINE 97155085
PUBMED 9001799
REMARK VARIANT CAIS CYS-855.
REFERENCE 88 (residues 1 to 919)
AUTHORS Bevan,C.L., Brown,B.B., Davies,H.R., Evans,B.A., Hughes,I.A. and Patterson,M.N.
TITLE Functional analysis of six androgen receptor mutations identified in patients with partial androgen insensitivity syndrome
JOURNAL Hum. Mol. Genet. 5 (2), 265-273 (1996)

MEDLINE 96422266
PUBMED 8824883
REMARK VARIANTS PAIS ILE-742; ILE-780; GLU-798; CYS-840; HIS-855 AND MET-869.
REFERENCE 89 (residues 1 to 919)
AUTHORS Choong,C.S., Sturm,M.J., Strophair,J.A., McCulloch,R.K., Tilley,W.D., Leedman,P.J. and Hurley,D.M.
TITLE Partial androgen insensitivity caused by an androgen receptor mutation at amino acid 907 (Gly-->Arg) that results in decreased ligand binding affinity and reduced androgen receptor messenger ribonucleic acid levels
JOURNAL J. Clin. Endocrinol. Metab. 81 (1), 236-243 (1996)
MEDLINE 96141994
PUBMED 8550758
REMARK VARIANT PAIS ARG-909.
REFERENCE 90 (residues 1 to 919)
AUTHORS Lumbroso,S., Lobaccaro,J.M., Georget,V., Leger,J., Poujol,N., Terouanne,B., Evain-Brion,D., Czernichow,P. and Sultan,C.
TITLE A novel substitution (Leu707Arg) in exon 4 of the androgen receptor gene causes complete androgen resistance
JOURNAL J. Clin. Endocrinol. Metab. 81 (5), 1984-1988 (1996)
MEDLINE 96210890
PUBMED 8626869
REMARK VARIANT CAIS ARG-707.
REFERENCE 91 (residues 1 to 919)
AUTHORS Rodien,P., Mebarki,F., Mowszowicz,I., Chaussain,J.L., Young,J., Morel,Y. and Schaison,G.
TITLE Different phenotypes in a family with androgen insensitivity caused by the same M780I point mutation in the androgen receptor gene
JOURNAL J. Clin. Endocrinol. Metab. 81 (8), 2994-2998 (1996)
MEDLINE 96320094
PUBMED 8768864
REMARK VARIANT PAIS/CAIS ILE-780.
REFERENCE 92 (residues 1 to 919)
AUTHORS Choong,C.S., Quigley,C.A., French,F.S. and Wilson,E.M.
TITLE A novel missense mutation in the amino-terminal domain of the human androgen receptor gene in a family with partial androgen insensitivity syndrome causes reduced efficiency of protein translation
JOURNAL J. Clin. Invest. 98 (6), 1423-1431 (1996)
MEDLINE 96420610
PUBMED 8823308
REMARK VARIANT PAIS LYS-2.
REFERENCE 93 (residues 1 to 919)
AUTHORS Bruggenwirth,H.T., Boehmer,A.L., Verleun-Mooijman,M.C., Hoogenboezem,T., Kleijer,W.J., Otten,B.J., Trapman,J. and Brinkmann,A.O.
TITLE Molecular basis of androgen insensitivity
JOURNAL J. Steroid Biochem. Mol. Biol. 58 (5-6), 569-575 (1996)
MEDLINE 97078859
PUBMED 8918984
REMARK VARIANT CAIS ASP-573.
REFERENCE 94 (residues 1 to 919)
AUTHORS Sutherland,R.W., Wiener,J.S., Hicks,J.P., Marcelli,M., Gonzales,E.T. Jr., Roth,D.R. and Lamb,D.J.
TITLE Androgen receptor gene mutations are rarely associated with isolated penile hypospadias
JOURNAL J. Urol. 156 (2 Pt 2), 828-831 (1996)
MEDLINE 96289452
PUBMED 8683794

REMARK VARIANT MAIS SER-548.
REFERENCE 95 (residues 1 to 919)
AUTHORS Lobaccaro,J.M., Poujol,N., Chiche,L., Lumbroso,S., Brown,T.R. and Sultan,C.
TITLE Molecular modeling and in vitro investigations of the human androgen receptor DNA-binding domain: application for the study of two mutations
JOURNAL Mol. Cell. Endocrinol. 116 (2), 137-147 (1996)
MEDLINE 96231386
PUBMED 8647313
REMARK VARIANT CAIS PRO-616.
REFERENCE 96 (residues 1 to 919)
AUTHORS Imasaki,K., Okabe,T., Murakami,H., Tanaka,Y., Haji,M., Takayanagi,R. and Nawata,H.
TITLE Androgen insensitivity syndrome due to new mutations in the DNA-binding domain of the androgen receptor
JOURNAL Mol. Cell. Endocrinol. 120 (1), 15-24 (1996)
MEDLINE 96405605
PUBMED 8809734
REMARK VARIANT CAIS PHE-579, AND VARIANT PAIS TYR-582.
REFERENCE 97 (residues 1 to 919)
AUTHORS Evans,B.A., Harper,M.E., Daniells,C.E., Watts,C.E., Matenhelia,S., Green,J. and Griffiths,K.
TITLE Low incidence of androgen receptor gene mutations in human prostatic tumors using single strand conformation polymorphism analysis
JOURNAL Prostate 28 (3), 162-171 (1996)
MEDLINE 96235947
PUBMED 8628719
REMARK VARIANT PROSTATE CANCER GLU-798.
REFERENCE 98 (residues 1 to 919)
AUTHORS Suzuki,H., Akakura,K., Komiya,A., Aida,S., Akimoto,S. and Shimazaki,J.
TITLE Codon 877 mutation in the androgen receptor gene in advanced prostate cancer: relation to antiandrogen withdrawal syndrome
JOURNAL Prostate 29 (3), 153-158 (1996)
MEDLINE 96424622
PUBMED 8827083
REMARK VARIANT PROSTATE CANCER ALA-877.
REFERENCE 99 (residues 1 to 919)
AUTHORS Boehmer,A.L., Brinkmann,A.O., Niermeijer,M.F., Bakker,L., Halley,D.J. and Drop,S.L.
TITLE Germ-line and somatic mosaicism in the androgen insensitivity syndrome: implications for genetic counseling
JOURNAL Am. J. Hum. Genet. 60 (4), 1003-1006 (1997)
MEDLINE 97260430
PUBMED 9106550
REMARK VARIANT PAIS HIS-855.
REFERENCE 100 (residues 1 to 919)
AUTHORS Koivisto,P., Kononen,J., Palmberg,C., Tammela,T., Hyytinen,E., Isola,J., Trapman,J., Cleutjens,K., Noordzij,A., Visakorpi,T. and Kallioniemi,O.-P.
TITLE Androgen receptor gene amplification: a possible molecular mechanism for androgen deprivation therapy failure in prostate cancer
JOURNAL Cancer Res. 57 (2), 314-319 (1997)
MEDLINE 97153285
PUBMED 9000575
REMARK VARIANT PROSTATE CANCER ALA-683.
REFERENCE 101 (residues 1 to 919)

AUTHORS Tincello,D.G., Saunders,P.T., Hodgins,M.B., Simpson,N.B., Edwards,C.R., Hargreaves,T.B. and Wu,F.C.
TITLE Correlation of clinical, endocrine and molecular abnormalities with in vivo responses to high-dose testosterone in patients with partial androgen insensitivity syndrome
JOURNAL Clin. Endocrinol. (Oxf) 46 (4), 497-506 (1997)
MEDLINE 97340104
PUBMED 9196614
REMARK VARIANTS PAIS LYS-608 AND GLY-772.
REFERENCE 102 (residues 1 to 919)
AUTHORS Essawi,M., Gad,Y.Z., el-Rouby,O., Temtamy,S.A., Sabour,Y.A. and el-Awady,M.K.
TITLE Molecular analysis of androgen resistance syndromes in Egyptian patients
JOURNAL Dis. Markers 13 (2), 99-105 (1997)
MEDLINE 97303883
PUBMED 9160185
REMARK VARIANT PAIS MET-889.
REFERENCE 103 (residues 1 to 919)
AUTHORS Sinnecker,G.H., Hiort,O., Nitsche,E.M., Holterhus,P.M. and Kruse,K.
TITLE Functional assessment and clinical classification of androgen sensitivity in patients with mutations of the androgen receptor gene. German Collaborative Intersex Study Group
JOURNAL Eur. J. Pediatr. 156 (1), 7-14 (1997)
MEDLINE 97159966
PUBMED 9007482
REMARK VARIANT CAIS TRP-779.
REFERENCE 104 (residues 1 to 919)
AUTHORS Jakubiczka,S., Nedel,S., Werder,E.A., Schleiermacher,E., Theile,U., Wolff,G. and Wieacker,P.
TITLE Mutations of the androgen receptor gene in patients with complete androgen insensitivity
JOURNAL Hum. Mutat. 9 (1), 57-61 (1997)
MEDLINE 97144184
PUBMED 8990010
REMARK VARIANTS CAIS VAL-749; CYS-774; ILE-780 AND SER-794.
REFERENCE 105 (residues 1 to 919)
AUTHORS Watanabe,M., Ushijima,T., Shiraishi,T., Yatani,R., Shimazaki,J., Kotake,T., Sugimura,T. and Nagao,M.
TITLE Genetic alterations of androgen receptor gene in Japanese human prostate cancer
JOURNAL Jpn J Clin Oncol 27 (6), 389-393 (1997)
MEDLINE 98100812
PUBMED 9438000
REMARK VARIANTS PROSTATE CANCER IN POLY-GLN REGION; HIS-701 AND ARG-910.
REFERENCE 106 (residues 1 to 919)
AUTHORS Wang,C. and Uchida,T.
TITLE Androgen receptor gene mutations in prostate cancer
JOURNAL Jpn. J. Urol. 88, 550-556 (1997)
REMARK VARIANT PROSTATE CANCER GLN-629.
REFERENCE 107 (residues 1 to 919)
AUTHORS Komori,S., Sakata,K., Tanaka,H., Shima,H. and Koyama,K.
TITLE DNA analysis of the androgen receptor gene in two cases with complete androgen insensitivity syndrome
JOURNAL J Obstet Gynaecol Res 23 (3), 277-281 (1997)
MEDLINE 97398857
PUBMED 9255042
REMARK VARIANTS CAIS ARG-194 AND CYS-855.
REFERENCE 108 (residues 1 to 919)
AUTHORS Albers,N., Ulrichs,C., Gluer,S., Hiort,O., Sinnecker,G.H.,

Mildenberger,H. and Brodehl,J.
TITLE Etiologic classification of severe hypospadias: implications for prognosis and management
JOURNAL J. Pediatr. 131 (3), 386-392 (1997)
MEDLINE 97469997
PUBMED 9329414
REMARK VARIANTS PAIS ALA-708 AND GLY-870.
REFERENCE 109 (residues 1 to 919)
AUTHORS Ko,T.M., Yang,Y.S., Wu,M.Y., Kao,C.H., Hsu,P.M., Chuang,S.M. and Lee,T.Y.
TITLE Complete androgen insensitivity syndrome. Molecular characterization in two Chinese women
JOURNAL J Reprod Med 42 (7), 424-428 (1997)
MEDLINE 97396769
PUBMED 9252933
REMARK VARIANTS CAIS ASN-732 AND THR-765.
REFERENCE 110 (residues 1 to 919)
AUTHORS Bevan,C.L., Hughes,I.A. and Patterson,M.N.
TITLE Wide variation in androgen receptor dysfunction in complete androgen insensitivity syndrome
JOURNAL J. Steroid Biochem. Mol. Biol. 61 (1-2), 19-26 (1997)
MEDLINE 97466866
PUBMED 9328206
REMARK VARIANTS CAIS ASP-750; PHE-762; THR-765; ASN-864 AND PHE-907.
REFERENCE 111 (residues 1 to 919)
AUTHORS Radmayr,C., Culig,Z., Glatzl,J., Neuschmid-Kaspar,F., Bartsch,G. and Klocker,H.
TITLE Androgen receptor point mutations as the underlying molecular defect in 2 patients with androgen insensitivity syndrome
JOURNAL J. Urol. 158 (4), 1553-1556 (1997)
MEDLINE 97445885
PUBMED 9302173
REMARK VARIANT PAIS GLY-703, AND VARIANT CAIS LEU-916.
REFERENCE 112 (residues 1 to 919)
AUTHORS Komori,S., Kasumi,H., Sakata,K., Tanaka,H., Hamada,K. and Koyama,K.
TITLE Molecular analysis of the androgen receptor gene in 4 patients with complete androgen insensitivity
JOURNAL Arch. Gynecol. Obstet. 261 (2), 95-100 (1998)
MEDLINE 98206019
PUBMED 9544375
REMARK VARIANTS CAIS CYS-571; GLN-752 AND CYS-774.
REFERENCE 113 (residues 1 to 919)
AUTHORS Cabral,D.F., Maciel-Guerra,A.T. and Hackel,C.
TITLE Mutations of androgen receptor gene in Brazilian patients with male pseudohermaphroditism
JOURNAL Braz. J. Med. Biol. Res. 31 (6), 775-778 (1998)
MEDLINE 98363978
PUBMED 9698822
REMARK VARIANTS CAIS HIS-615 AND GLN-752.
REFERENCE 114 (residues 1 to 919)
AUTHORS Wang,Q., Ghadessy,F.J. and Yong,E.L.
TITLE Analysis of the transactivation domain of the androgen receptor in patients with male infertility
JOURNAL Clin. Genet. 54 (3), 185-192 (1998)
MEDLINE 99002768
PUBMED 9788719
REMARK VARIANT MAIS ARG-214.
REFERENCE 115 (residues 1 to 919)
AUTHORS Tanaka,H., Komori,S., Sakata,K., Shima,H. and Koyama,K.
TITLE One additional mutation at exon A amplifies thermolability of

androgen receptor in a case with complete androgen insensitivity syndrome
JOURNAL Gynecol Endocrinol 12 (2), 75-82 (1998)
MEDLINE 98273339
PUBMED 9610419
REMARK VARIANTS CAIS PRO-255 AND ALA-820.
REFERENCE 116 (residues 1 to 919)
AUTHORS Lundberg Giwercman,Y., Nikoshkov,A., Lindsten,K., Bystroem,B., Pousette,A., Chibalin,A.V., Arvidsson,S., Tiulpakov,A., Semitcheva,T.V., Peterkova,V., Hagenfeldt,K., Ritzen,E.M. and Wedell,A.
TITLE Functional characterisation of mutations in the ligand-binding domain of the androgen receptor gene in patients with androgen insensitivity syndrome
JOURNAL Hum. Genet. 103 (4), 529-531 (1998)
MEDLINE 99072324
PUBMED 9856504
REMARK VARIANTS CAIS THR-765; TYR-784 AND THR-895, AND VARIANT PAIS GLY-840.
REFERENCE 117 (residues 1 to 919)
AUTHORS Doerk,T., Schnieders,F., Jakubiczka,S., Wieacker,P., Schroeder-Kurth,T. and Schmidtke,J.
TITLE A new missense substitution at a mutational hot spot of the androgen receptor in siblings with complete androgen insensitivity syndrome
JOURNAL Hum. Mutat. 11, 337-339 (1998)
REMARK VARIANT CAIS VAL-695.
REFERENCE 118 (residues 1 to 919)
AUTHORS Nordenskjoeld,A. and Soederhaell,S.
TITLE An androgen receptor gene mutation (A645D) in a boy with a normal phenotype
JOURNAL Hum. Mutat. 11, 339-339 (1998)
REMARK VARIANT ASP-645.
REFERENCE 119 (residues 1 to 919)
AUTHORS Knoke,I., Jakubiczka,S., Rohrer,T., Hanimann,B., Werder,E.A. and Wieacker,P.
TITLE Single amino acid substitution in the hormone-binding domain of the androgen receptor in a family with complete androgen insensitivity syndrome (CAIS)
JOURNAL Hum. Mutat. 12, 220-220 (1998)
REMARK VARIANT CAIS LEU-892.
REFERENCE 120 (residues 1 to 919)
AUTHORS Weidemann,W., Peters,B., Romalo,G., Spindler,K.D. and Schweikert,H.U.
TITLE Response to androgen treatment in a patient with partial androgen insensitivity and a mutation in the deoxyribonucleic acid-binding domain of the androgen receptor
JOURNAL J. Clin. Endocrinol. Metab. 83 (4), 1173-1176 (1998)
MEDLINE 98202136
PUBMED 9543136
REMARK VARIANT PAIS GLN-607.
REFERENCE 121 (residues 1 to 919)
AUTHORS Georget,V., Terouanne,B., Lumbroso,S., Nicolas,J.C. and Sultan,C.
TITLE Trafficking of androgen receptor mutants fused to green fluorescent protein: a new investigation of partial androgen insensitivity syndrome
JOURNAL J. Clin. Endocrinol. Metab. 83 (10), 3597-3603 (1998)
MEDLINE 98439607
PUBMED 9768671
REMARK VARIANTS PAIS VAL-743 AND CYS-840.

REFERENCE 122 (residues 1 to 919)
AUTHORS Wang,Q., Ghadessy,F.J., Trounson,A., de Kretser,D., McLachlan,R., Ng,S.C. and Yong,E.L.
TITLE Azoospermia associated with a mutation in the ligand-binding domain of an androgen receptor displaying normal ligand binding, but defective trans-activation
JOURNAL J. Clin. Endocrinol. Metab. 83 (12), 4303-4309 (1998)
MEDLINE 99067093
PUBMED 9851768
REMARK VARIANT MAIS GLU-798.
REFERENCE 123 (residues 1 to 919)
AUTHORS Hiort,O., Sinnecker,G.H., Holterhus,P.M., Nitsche,E.M. and Kruse,K.
TITLE Inherited and de novo androgen receptor gene mutations: investigation of single-case families
JOURNAL J. Pediatr. 132 (6), 939-943 (1998)
MEDLINE 98291052
PUBMED 9627582
REMARK VARIANTS CAIS AND PAIS.
REFERENCE 124 (residues 1 to 919)
AUTHORS Yong,E.L., Tut,T.G., Ghadessy,F.J., Prins,G. and Ratnam,S.S.
TITLE Partial androgen insensitivity and correlations with the predicted three dimensional structure of the androgen receptor ligand-binding domain
JOURNAL Mol. Cell. Endocrinol. 137 (1), 41-50 (1998)
MEDLINE 98268743
PUBMED 9607727
REMARK VARIANT PAIS THR-758.
REFERENCE 125 (residues 1 to 919)
AUTHORS Knoke,I., Jakubiczka,S., Lehnert,H. and Wieacker,P.
TITLE A new point mutation of the androgen receptor gene in a patient with partial androgen resistance and severe oligozoospermia
JOURNAL Andrologia 31 (4), 199-201 (1999)
MEDLINE 99399430
PUBMED 10470409
REMARK VARIANT PAIS LEU-911.
REFERENCE 126 (residues 1 to 919)
AUTHORS Taplin,M.E., Bubley,G.J., Ko,Y.J., Small,E.J., Upton,M., Rajeshkumar,B. and Balk,S.P.
TITLE Selection for androgen receptor mutations in prostate cancers treated with androgen antagonist
JOURNAL Cancer Res. 59 (11), 2511-2515 (1999)
MEDLINE 99290631
PUBMED 10363963
REMARK VARIANTS PROSTATE CANCER ALA-877 AND ASN-890.
REFERENCE 127 (residues 1 to 919)
AUTHORS Melo,K.F.S., Latronico,A.C., Costa,E.M.F., Billerbeck,A.E.C., Mendonca,B.B. and Arnhold,I.J.P.
TITLE A novel point mutation (R840S) in the androgen receptor in a Brazilian family with partial androgen insensitivity syndrome
JOURNAL Hum. Mutat. 14, 353-353 (1999)
REMARK VARIANT PAIS SER-840.
REFERENCE 128 (residues 1 to 919)
AUTHORS Gottlieb,B., Vasiliou,D.M., Lumbroso,R., Beitel,L.K., Pinsky,L. and Trifiro,M.A.
TITLE Analysis of exon 1 mutations in the androgen receptor gene
JOURNAL Hum. Mutat. 14 (6), 527-539 (1999)
MEDLINE 20040031
PUBMED 10571951
REMARK VARIANTS CAIS ARG-390 AND ARG-443.
REFERENCE 129 (residues 1 to 919)

AUTHORS Chen,C.P., Chern,S.R., Wang,T.Y., Wang,W., Wang,K.L. and Jeng,C.J.
TITLE Androgen receptor gene mutations in 46,XY females with germ cell tumours
JOURNAL Hum. Reprod. 14 (3), 664-670 (1999)
MEDLINE 99236881
PUBMED 10221692
REMARK VARIANT PAIS GLN-607, AND VARIANT CAIS LYS-681.
REFERENCE 130 (residues 1 to 919)
AUTHORS Kanayama,H., Naroda,T., Inoue,Y., Kurokawa,Y. and Kagawa,S.
TITLE A case of complete testicular feminization: laparoscopic orchiectomy and analysis of androgen receptor gene mutation
JOURNAL Int J Urol 6 (6), 327-330 (1999)
MEDLINE 99332382
PUBMED 10404311
REMARK VARIANT CAIS LEU-892.
REFERENCE 131 (residues 1 to 919)
AUTHORS Shkolny,D.L., Beitel,L.K., Ginsberg,J., Pekeles,G., Arbour,L., Pinsky,L. and Trifiro,M.A.
TITLE Discordant measures of androgen-binding kinetics in two mutant androgen receptors causing mild or partial androgen insensitivity, respectively
JOURNAL J. Clin. Endocrinol. Metab. 84 (2), 805-810 (1999)
MEDLINE 99145056
PUBMED 10022458
REMARK VARIANT PAIS ALA-772, AND VARIANT MAIS GLY-871.
REFERENCE 132 (residues 1 to 919)
AUTHORS Wallen,M.J., Linja,M., Kaartinen,K., Schleutker,J. and Visakorpi,T.
TITLE Androgen receptor gene mutations in hormone-refractory prostate cancer
JOURNAL J. Pathol. 189 (4), 559-563 (1999)
MEDLINE 20096941
PUBMED 10629558
REMARK VARIANTS PROSTATE CANCER IN POLY-GLN REGION AND ALA-683.
REFERENCE 133 (residues 1 to 919)
AUTHORS Zhao,X.Y., Boyle,B., Krishnan,A.V., Navone,N.M., Peehl,D.M. and Feldman,D.
TITLE Two mutations identified in the androgen receptor of the new human prostate cancer cell line MDA PCa 2a
JOURNAL J. Urol. 162 (6), 2192-2199 (1999)
MEDLINE 20034906
PUBMED 10569618
REMARK VARIANTS PROSTATE CANCER HIS-701 AND ALA-877.
REFERENCE 134 (residues 1 to 919)
AUTHORS Ong,Y.C., Wong,H.B., Adaikan,G. and Yong,E.L.
TITLE Directed pharmacological therapy of ambiguous genitalia due to an androgen receptor gene mutation
JOURNAL Lancet 354 (9188), 1444-1445 (1999)
MEDLINE 20009021
PUBMED 10543676
REMARK VARIANT PAIS THR-807.
REFERENCE 135 (residues 1 to 919)
AUTHORS Peters,I., Weidemann,W., Romalo,G., Knorr,D., Schweikert,H.U. and Spindler,K.D.
TITLE An androgen receptor mutation in the direct vicinity of the proposed C-terminal alpha-helix of the ligand binding domain containing the AF-2 transcriptional activating function core is associated with complete androgen insensitivity
JOURNAL Mol. Cell. Endocrinol. 148 (1-2), 47-53 (1999)
MEDLINE 99236959
PUBMED 10221770

REMARK VARIANT CAIS LEU-892.
REFERENCE 136 (residues 1 to 919)
AUTHORS Nazareth,L.V., Stenoien,D.L., Bingman,W.E. III, James,A.J., Wu,C., Zhang,Y., Edwards,D.P., Mancini,M., Marcelli,M., Lamb,D.J. and Weigel,N.L.
TITLE A C619Y mutation in the human androgen receptor causes inactivation and mislocalization of the receptor with concomitant sequestration of SRC-1 (steroid receptor coactivator 1)
JOURNAL Mol. Endocrinol. 13 (12), 2065-2075 (1999)
MEDLINE 20065641
PUBMED 10598582
REMARK VARIANT PROSTATE CANCER TYR-619.
REFERENCE 137 (residues 1 to 919)
AUTHORS Nazareth,L.V., Stenoien,D.L., Bingman,W.E. III, James,A.J., Wu,C., Zhang,Y., Edwards,D.P., Mancini,M., Marcelli,M., Lamb,D.J. and Weigel,N.L.
JOURNAL Mol. Endocrinol. 14, 544-544 (2000)
REMARK ERRATUM.
REFERENCE 138 (residues 1 to 919)
AUTHORS Holterhus,P.M., Wiebel,J., Sinnecker,G.H., Bruggenwirth,H.T., Sippell,W.G., Brinkmann,A.O., Kruse,K. and Hiort,O.
TITLE Clinical and molecular spectrum of somatic mosaicism in androgen insensitivity syndrome
JOURNAL Pediatr. Res. 46 (6), 684-690 (1999)
MEDLINE 20055969
PUBMED 10590024
REMARK VARIANT PAIS THR-596.
REFERENCE 139 (residues 1 to 919)
AUTHORS Yaegashi,N., Uehara,S., Senoo,M., Sato,J., Fujiwara,J., Funato,T., Sasaki,T. and Yajima,A.
TITLE Point mutations in the steroid-binding domain of the androgen receptor gene of five Japanese patients with androgen insensitivity syndrome
JOURNAL Tohoku J. Exp. Med. 187 (3), 263-272 (1999)
MEDLINE 99385665
PUBMED 10458483
REMARK VARIANT PAIS PHE-812, AND VARIANT CAIS GLN-831.
REFERENCE 140 (residues 1 to 919)
AUTHORS Nordenskjold,A., Friedman,E., Tapper-Persson,M., Soderhall,C., Leviav,A., Svensson,J. and Anvret,M.
TITLE Screening for mutations in candidate genes for hypospadias
JOURNAL Urol. Res. 27 (1), 49-55 (1999)
MEDLINE 99190574
PUBMED 10092153
REMARK VARIANTS PAIS THR-597 AND LEU-725.
REFERENCE 141 (residues 1 to 919)
AUTHORS Marcelli,M., Ittmann,M., Mariani,S., Sutherland,R.W., Nigam,R., Murthy,L., Zhao,Y., DiConcini,D., Puxeddu,E., Esen,A., Eastham,J., Weigel,N.L. and Lamb,D.J.
TITLE Androgen receptor mutations in prostate cancer
JOURNAL Cancer Res. 60 (4), 944-949 (2000)
MEDLINE 20168626
PUBMED 10706109
REMARK VARIANTS PROSTATE CANCER ALA-575; ARG-580; VAL-586; TYR-619; ALA-757 AND GLY-846.
REFERENCE 142 (residues 1 to 919)
AUTHORS Ahmed,S.F., Cheng,A., Dovey,L., Hawkins,J.R., Martin,H., Rowland,J., Shimura,N., Tait,A.D. and Hughes,I.A.
TITLE Phenotypic features, androgen receptor binding, and mutational analysis in 278 clinical cases reported as androgen insensitivity

syndrome
JOURNAL J. Clin. Endocrinol. Metab. 85 (2), 658-665 (2000)
MEDLINE 20152731
PUBMED 10690872
REMARK VARIANTS CAIS AND PAIS.
REFERENCE 143 (residues 1 to 919)
AUTHORS Chavez,B., Mendez,J.P., Ulloa-Aguirre,A., Larrea,F. and Vilchis,F.
TITLE Eight novel mutations of the androgen receptor gene in patients with androgen insensitivity syndrome
JOURNAL J. Hum. Genet. 46 (10), 560-565 (2001)
MEDLINE 21470699
PUBMED 11587068
REMARK VARIANTS PAIS THR-682 AND GLU-711, VARIANTS CAIS GLU-743; VAL-827 AND ARG-874, AND VARIANT MAIS TYR-879.
REFERENCE 144 (residues 1 to 919)
AUTHORS Sills,E.S., Sholes,T.E., Perloe,M., Kaplan,C.R., Davis,J.G. and Tucker,M.J.
TITLE Characterization of a novel receptor mutation A-->T at exon 4 in complete androgen insensitivity syndrome and a carrier sibling via bidirectional polymorphism sequence analysis
JOURNAL Int. J. Mol. Med. 9 (1), 45-48 (2002)
MEDLINE 21610990
PUBMED 11744994
REMARK VARIANT CAIS TYR-705.
COMMENT ----------

[FUNCTION] THE STEROID HORMONES AND THEIR RECEPTORS ARE INVOLVED IN THE REGULATION OF EUKARYOTIC GENE EXPRESSION AND AFFECT CELLULAR PROLIFERATION AND DIFFERENTIATION IN TARGET TISSUES.
[SUBUNIT] Binds DNA as a homodimer. The AR N-terminal poly-Gln region binds RAN resulting in enhancement of AR-mediated transactivation. RAN binding decreases as the poly-Gln length increases.
[SUBCELLULAR LOCATION] Nuclear.
[DOMAIN] Composed of three domains: a modulating N-terminal domain, a DNA-binding domain and a C-terminal steroid-binding domain.
[POLYMORPHISM] The poly-Gln region of AR is highly polymorphic and the number of Gln varies in the population (from 17 to 26). A smaller size of the poly-Gln region may be associated with the development of prostate cancer.
[POLYMORPHISM] The poly-Gly region of AR is also polymorphic and ranges from 24 to 31 Gly.
[DISEASE] DEFECTS IN AR ARE THE CAUSE OF ANDROGEN INSENSIBILITY SYNDROME (AIS), PREVIOUSLY KNOWN AS TESTICULAR FEMINIZATION SYNDROME (TFM). IT CAN BE COMPLETE (CAIS) WHEN EXTERNAL GENITALIA ARE PHENOTYPICALLY FEMALE; OR PARTIAL (PAIS) WHEN EXTERNAL GENITALIA ARE SUBSTANTIVELY AMBIGUOUS OR MILD (MAIS) WHEN EXTERNAL GENITALIA ARE NORMAL MALE OR NEARLY SO.
[DISEASE] DEFECTS IN AR ARE THE CAUSE OF X-LINKED SPINAL AND BULBAR MUSCULAR ATROPHY (SBMA) (ALSO KNOWN AS KENNEDY'S DISEASE). IN SBMA PATIENTS THE NUMBER OF GLN RANGES FROM 40 TO 52. LONGER EXPANSIONS RESULT IN EARLIER ONSET AND MORE SEVERE CLINICAL MANIFESTATIONS OF THE DISEASE.
[DISEASE] DEFECTS IN AR MAY PLAY A ROLE IN METASTATIC PROSTATE CANCER. THE MUTATED RECEPTOR STIMULATES PROSTATE GROWTH AND

```
            METASTASES DEVELOPMENT DESPITE OF ANDROGEN ABLATION. THIS TREATMENT
            CAN REDUCE PRIMARY AND METASTATIC LESIONS PROBABLY BY INDUCING
            APOPTOSIS OF TUMOR CELLS WHEN THEY EXPRESS THE WILD-TYPE RECEPTOR.
            [DISEASE] DEFECTS IN AR MAY BE THE CAUSE OF INFERTILITY MALE
            SYNDROME. IT IS CHARACTERIZED BY AZOOSPERMIA, ELEVATED TESTOSTERONE
            AND LUTEINIZING HORMONE PLASMA LEVELS AND AN ABNORMAL ANDROGEN
            RECEPTOR.
            [MISCELLANEOUS] In the absence of ligand, steroid hormone receptors
            are thought to be weakly associated with nuclear components;
            hormone binding greatly increases receptor affinity. The
            hormone-receptor complex appears to recognize discrete DNA
            sequences upstream of transcriptional start sites.
            [SIMILARITY] Belongs to the nuclear hormone receptor family. NR3
            subfamily.
            [DATABASE] NAME=Androgen receptor gene mutations database;
            WWW='http://www.mcgill.ca/androgendb/';
            FTP='ftp://ftp.ebi.ac.uk/pub/databases/androgen'.
FEATURES             Location/Qualifiers
     source          1..919
                     /organism="Homo sapiens"
                     /mol_type="unassigned DNA"
                     /db_xref="taxon:9606"
     gene            1..919
                     /gene="AR"
                     /note="synonyms: NR3C4, DHTR"
     Protein         1..919
                     /gene="AR"
                     /product="Androgen receptor"
     Region          1..558
                     /gene="AR"
                     /region_name="Domain"
                     /note="MODULATING."
     Region          2
                     /gene="AR"
                     /region_name="Variant"
                     /note="E -> K (IN PAIS). /FTId=VAR_004679."
     Region          54..57
                     /gene="AR"
                     /region_name="Domain"
                     /note="POLY-LEU."
     Region          54
                     /gene="AR"
                     /region_name="Variant"
                     /note="L -> S (IN PROSTATE CANCER). /FTId=VAR_004680."
     Region          57
                     /gene="AR"
                     /region_name="Variant"
                     /note="L -> Q (IN PROSTATE CANCER). /FTId=VAR_004681."
     Region          58..89
                     /gene="AR"
                     /region_name="Domain"
                     /note="GLN-RICH."
     Region          58..78
                     /gene="AR"
                     /region_name="Domain"
                     /note="POLY-GLN."
     Region          64
                     /gene="AR"
                     /region_name="Variant"
                     /note="Q -> R (IN PROSTATE CANCER). /FTId=VAR_009711."
```

```
Region          74..78
                /gene="AR"
                /region_name="Variant"
                /note="MISSING. /FTId=VAR_004682."
Region          84..89
                /gene="AR"
                /region_name="Domain"
                /note="POLY-GLN."
Region          112
                /gene="AR"
                /region_name="Variant"
                /note="Q -> H (IN PROSTATE CANCER). /FTId=VAR_009712."
Region          166
                /gene="AR"
                /region_name="Conflict"
                /note="G -> A (IN REF. 3)."
Region          180
                /gene="AR"
                /region_name="Variant"
                /note="K -> R (IN PROSTATE CANCER). /FTId=VAR_009713."
Region          193..197
                /gene="AR"
                /region_name="Domain"
                /note="POLY-GLN."
Region          194
                /gene="AR"
                /region_name="Variant"
                /note="Q -> R (IN CAIS). /FTId=VAR_009224."
Region          205
                /gene="AR"
                /region_name="Variant"
                /note="S -> R. /FTId=VAR_009714."
Region          212
                /gene="AR"
                /region_name="Conflict"
                /note="A -> R (IN REF. 5 AND 6)."
Region          214
                /gene="AR"
                /region_name="Variant"
                /note="G -> R (IN MAIS; 20% LOWER TRANSACTIVATION
                CAPACITY). /FTId=VAR_009715."
Region          255
                /gene="AR"
                /region_name="Variant"
                /note="L -> P (IN CAIS). /FTId=VAR_009225."
Region          266
                /gene="AR"
                /region_name="Variant"
                /note="M -> T (IN PROSTATE CANCER). /FTId=VAR_009716."
Region          269
                /gene="AR"
                /region_name="Variant"
                /note="P -> S (IN PROSTATE CANCER). /FTId=VAR_009717."
Region          340
                /gene="AR"
                /region_name="Variant"
                /note="P -> L (IN PROSTATE CANCER). /FTId=VAR_009718."
Region          372..381
                /gene="AR"
                /region_name="Domain"
```

```
                /note="POLY-PRO."
     Region     390
                /gene="AR"
                /region_name="Variant"
                /note="P -> R (IN CAIS). /FTId=VAR_009226."
     Region     390
                /gene="AR"
                /region_name="Variant"
                /note="P -> S (IN MAIS). /FTId=VAR_009227."
     Region     396..402
                /gene="AR"
                /region_name="Domain"
                /note="POLY-ALA."
     Region     443
                /gene="AR"
                /region_name="Variant"
                /note="Q -> R (IN CAIS; MIGHT BE A POLYMORPHISM).
                /FTId=VAR_009228."
     Region     449..472
                /gene="AR"
                /region_name="Domain"
                /note="POLY-GLY."
     Region     465..472
                /gene="AR"
                /region_name="Variant"
                /note="MISSING. /FTId=VAR_004683."
     Region     475
                /gene="AR"
                /region_name="Conflict"
                /note="G -> E (IN REF. 4 AND 7)."
     Region     491
                /gene="AR"
                /region_name="Variant"
                /note="G -> S (IN CAIS). /FTId=VAR_009719."
     Region     528
                /gene="AR"
                /region_name="Variant"
                /note="D -> G (IN PROSTATE CANCER). /FTId=VAR_009720."
     Region     547
                /gene="AR"
                /region_name="Variant"
                /note="L -> F (IN PAIS). /FTId=VAR_009721."
     Region     548
                /gene="AR"
                /region_name="Variant"
                /note="P -> S (IN MAIS). /FTId=VAR_009722."
     Site       559..624
                /gene="AR"
                /site_type="DNA binding"
                /note="NUCLEAR RECEPTOR-TYPE."
     Region     559..579
                /gene="AR"
                /region_name="Zinc finger region"
                /note="C4-TYPE."
     Region     559
                /gene="AR"
                /region_name="Variant"
                /note="C -> Y (IN CAIS). /FTId=VAR_009723."
     Region     565
                /gene="AR"
```

```
                /region_name="Conflict"
                /note="E -> K (IN REF. 8)."
     Region     568
                /gene="AR"
                /region_name="Variant"
                /note="G -> V (IN PAIS). /FTId=VAR_009725."
     Region     568
                /gene="AR"
                /region_name="Variant"
                /note="G -> W (IN PAIS). /FTId=VAR_009726."
     Region     571
                /gene="AR"
                /region_name="Variant"
                /note="Y -> C (IN CAIS). /FTId=VAR_009727."
     Region     573
                /gene="AR"
                /region_name="Variant"
                /note="A -> D (IN CAIS; DEFECTIVE DNA BINDING AND
                TRANSACTIVATION). /FTId=VAR_009728."
     Region     574
                /gene="AR"
                /region_name="Variant"
                /note="L -> P (IN PROSTATE CANCER). /FTId=VAR_009729."
     Region     575
                /gene="AR"
                /region_name="Variant"
                /note="T -> A (IN PROSTATE CANCER). /FTId=VAR_009730."
     Region     576
                /gene="AR"
                /region_name="Variant"
                /note="C -> F (IN CAIS; LACK OF DNA BINDING).
                /FTId=VAR_009731."
     Region     576
                /gene="AR"
                /region_name="Variant"
                /note="C -> R (IN CAIS). /FTId=VAR_009732."
     Region     579
                /gene="AR"
                /region_name="Variant"
                /note="C -> F (IN CAIS; REDUCED TRANSCRIPTION AND DNA
                BINDING). /FTId=VAR_009733."
     Region     579
                /gene="AR"
                /region_name="Variant"
                /note="C -> Y (IN CAIS). /FTId=VAR_009734."
     Region     580
                /gene="AR"
                /region_name="Variant"
                /note="K -> R (IN PROSTATE CANCER). /FTId=VAR_009735."
     Region     581
                /gene="AR"
                /region_name="Variant"
                /note="V -> F (IN CAIS). /FTId=VAR_009736."
     Region     582
                /gene="AR"
                /region_name="Variant"
                /note="F -> S (IN PAIS). /FTId=VAR_009737."
     Region     582
                /gene="AR"
                /region_name="Variant"
```

```
                /note="F -> Y (IN PAIS). /FTId=VAR_009738."
     Region     582
                /gene="AR"
                /region_name="Variant"
                /note="MISSING (IN CAIS). /FTId=VAR_009739."
     Region     585
                /gene="AR"
                /region_name="Variant"
                /note="R -> K (IN CAIS). /FTId=VAR_009740."
     Region     586
                /gene="AR"
                /region_name="Variant"
                /note="A -> V (IN PROSTATE CANCER; SOMATIC MUTATION).
                /FTId=VAR_009741."
     Region     587
                /gene="AR"
                /region_name="Variant"
                /note="A -> S (IN PROSTATE CANCER; SOMATIC MUTATION).
                /FTId=VAR_009742."
     Region     595..619
                /gene="AR"
                /region_name="Zinc finger region"
                /note="C4-TYPE."
     Region     596
                /gene="AR"
                /region_name="Variant"
                /note="A -> T (IN PAIS; ABOLISHES DIMERIZATION).
                /FTId=VAR_009743."
     Region     597
                /gene="AR"
                /region_name="Variant"
                /note="S -> G (IN PAIS; HIGH DISSOCIATION RATE; ASSOCIATED
                WITH P-617 IN A PAIS PATIENT; PARTIALLY RESTORES
                DNA-BINDING ACTIVITY OF P-617 MUTANT RECEPTORS).
                /FTId=VAR_009744."
     Region     597
                /gene="AR"
                /region_name="Variant"
                /note="S -> T (IN PAIS). /FTId=VAR_009745."
     Region     601
                /gene="AR"
                /region_name="Variant"
                /note="C -> F (IN CAIS). /FTId=VAR_009746."
     Region     604
                /gene="AR"
                /region_name="Variant"
                /note="D -> Y (IN PAIS). /FTId=VAR_009747."
     Region     607
                /gene="AR"
                /region_name="Variant"
                /note="R -> Q (IN PAIS AND BREAST CANCER).
                /FTId=VAR_004684."
     Region     608
                /gene="AR"
                /region_name="Variant"
                /note="R -> K (IN PAIS AND BREAST CANCER; DEFECTIVE
                NUCLEAR LOCALIZATION). /FTId=VAR_004685."
     Region     610
                /gene="AR"
                /region_name="Variant"
```

```
                /note="N -> T (IN PAIS). /FTId=VAR_009748."
     Region     611
                /gene="AR"
                /region_name="Variant"
                /note="C -> Y (IN CAIS). /FTId=VAR_009749."
     Region     615
                /gene="AR"
                /region_name="Variant"
                /note="MISSING (IN CAIS). /FTId=VAR_009750."
     Region     615
                /gene="AR"
                /region_name="Variant"
                /note="R -> H (IN CAIS AND PAIS). /FTId=VAR_009751."
     Region     615
                /gene="AR"
                /region_name="Variant"
                /note="R -> P (IN CAIS). /FTId=VAR_009752."
     Region     616
                /gene="AR"
                /region_name="Variant"
                /note="L -> P (IN CAIS). /FTId=VAR_009753."
     Region     616
                /gene="AR"
                /region_name="Variant"
                /note="L -> R (IN PAIS). /FTId=VAR_009754."
     Region     617
                /gene="AR"
                /region_name="Variant"
                /note="R -> P (IN CAIS AND PAIS; LOSS OF DNA- BINDING
                ACTIVITY; ASSOCIATED WITH G-597 IN A PAIS PATIENT).
                /FTId=VAR_009755."
     Region     619
                /gene="AR"
                /region_name="Variant"
                /note="C -> Y (IN PROSTATE CANCER; LOSS OF DNA BINDING;
                SOMATIC MUTATION). /FTId=VAR_009756."
     Region     629
                /gene="AR"
                /region_name="Variant"
                /note="R -> Q (IN PROSTATE CANCER). /FTId=VAR_009757."
     Region     630
                /gene="AR"
                /region_name="Variant"
                /note="K -> T (IN PROSTATE CANCER). /FTId=VAR_009758."
     Region     634
                /gene="AR"
                /region_name="Conflict"
                /note="L -> P (IN REF. 24; AAB21256/AAB21257)."
     Region     645
                /gene="AR"
                /region_name="Variant"
                /note="A -> D (IN dbSNP:1800053). /FTId=VAR_004686."
     Region     647
                /gene="AR"
                /region_name="Variant"
                /note="S -> N (IN PROSTATE CANCER). /FTId=VAR_009760."
     Region     664
                /gene="AR"
                /region_name="Variant"
                /note="I -> N (IN CAIS AND PAIS). /FTId=VAR_004687."
```

```
Region          670
                /gene="AR"
                /region_name="Variant"
                /note="Q -> R (IN PROSTATE CANCER). /FTId=VAR_009761."
Region          671
                /gene="AR"
                /region_name="Variant"
                /note="P -> H (IN PAIS). /FTId=VAR_009762."
Region          672
                /gene="AR"
                /region_name="Variant"
                /note="I -> T (IN PROSTATE CANCER). /FTId=VAR_009763."
Region          675
                /gene="AR"
                /region_name="Conflict"
                /note="N -> I (IN REF. 24; AAB21256/AAB21257)."
Region          677
                /gene="AR"
                /region_name="Variant"
                /note="L -> P (IN CAIS). /FTId=VAR_004688."
Region          681
                /gene="AR"
                /region_name="Variant"
                /note="E -> K (IN CAIS). /FTId=VAR_009764."
Region          682
                /gene="AR"
                /region_name="Variant"
                /note="P -> T (IN PAIS). /FTId=VAR_013474."
Region          683
                /gene="AR"
                /region_name="Variant"
                /note="G -> A (IN PROSTATE CANCER). /FTId=VAR_009765."
Region          684
                /gene="AR"
                /region_name="Variant"
                /note="V -> I (IN CAIS). /FTId=VAR_009766."
Region          686
                /gene="AR"
                /region_name="Variant"
                /note="C -> R (IN PAIS). /FTId=VAR_009767."
Region          687
                /gene="AR"
                /region_name="Variant"
                /note="A -> V (IN PAIS). /FTId=VAR_009768."
Region          688
                /gene="AR"
                /region_name="Variant"
                /note="G -> E (IN CAIS). /FTId=VAR_009769."
Region          690..919
                /gene="AR"
                /region_name="Domain"
                /note="LIGAND-BINDING."
Region          690
                /gene="AR"
                /region_name="Variant"
                /note="MISSING (IN PAIS). /FTId=VAR_009770."
Region          692
                /gene="AR"
                /region_name="Variant"
                /note="MISSING (IN CAIS). /FTId=VAR_004689."
```

```
Region          695
                /gene="AR"
                /region_name="Variant"
                /note="D -> H (IN CAIS). /FTId=VAR_004690."
Region          695
                /gene="AR"
                /region_name="Variant"
                /note="D -> N (IN CAIS). /FTId=VAR_004691."
Region          695
                /gene="AR"
                /region_name="Variant"
                /note="D -> V (IN CAIS). /FTId=VAR_004692."
Region          700
                /gene="AR"
                /region_name="Variant"
                /note="L -> M (IN CAIS). /FTId=VAR_009771."
Region          701
                /gene="AR"
                /region_name="Variant"
                /note="L -> F (IN CAIS)../FTId=VAR_009772."
Region          701
                /gene="AR"
                /region_name="Variant"
                /note="L -> H (IN CAIS AND PROSTATE CANCER).
                /FTId=VAR_009773."
Region          702
                /gene="AR"
                /region_name="Variant"
                /note="S -> A (IN CAIS). /FTId=VAR_009774."
Region          703
                /gene="AR"
                /region_name="Variant"
                /note="S -> C (IN CAIS). /FTId=VAR_009775."
Region          703
                /gene="AR"
                /region_name="Variant"
                /note="S -> G (IN PAIS AND CAIS). /FTId=VAR_004693."
Region          705
                /gene="AR"
                /region_name="Variant"
                /note="N -> S (IN CAIS). /FTId=VAR_009776."
Region          705
                /gene="AR"
                /region_name="Variant"
                /note="N -> Y (IN CAIS). /FTId=VAR_013475."
Region          707
                /gene="AR"
                /region_name="Variant"
                /note="L -> R (IN CAIS). /FTId=VAR_004694."
Region          708
                /gene="AR"
                /region_name="Variant"
                /note="G -> A (IN PAIS). /FTId=VAR_009777."
Region          708
                /gene="AR"
                /region_name="Variant"
                /note="G -> V (IN CAIS). /FTId=VAR_009778."
Region          710
                /gene="AR"
                /region_name="Variant"
```

```
                /note="R -> T (IN CAIS). /FTId=VAR_009779."
     Region     711
                /gene="AR"
                /region_name="Variant"
                /note="Q -> E (IN PAIS). /FTId=VAR_013476."
     Region     712
                /gene="AR"
                /region_name="Variant"
                /note="L -> F (IN PAIS). /FTId=VAR_009780."
     Region     715
                /gene="AR"
                /region_name="Variant"
                /note="V -> M (IN PROSTATE CANCER; GAIN IN FUNCTION).
                /FTId=VAR_009781."
     Region     717
                /gene="AR"
                /region_name="Variant"
                /note="K -> E (IN PROSTATE CANCER). /FTId=VAR_009782."
     Region     720
                /gene="AR"
                /region_name="Variant"
                /note="K -> E (IN PROSTATE CANCER; FOUND IN BONE
                METASTASES). /FTId=VAR_009783."
     Region     721
                /gene="AR"
                /region_name="Variant"
                /note="A -> T (IN PROSTATE CANCER; SOMATIC MUTATION).
                /FTId=VAR_009784."
     Region     722
                /gene="AR"
                /region_name="Variant"
                /note="L -> F (IN CAIS). /FTId=VAR_009785."
     Region     723
                /gene="AR"
                /region_name="Variant"
                /note="P -> S (IN CAIS). /FTId=VAR_009786."
     Region     724
                /gene="AR"
                /region_name="Variant"
                /note="G -> D (IN CAIS AND PROSTATE CANCER).
                /FTId=VAR_009787."
     Region     725
                /gene="AR"
                /region_name="Variant"
                /note="F -> L (IN PAIS). /FTId=VAR_009788."
     Region     726
                /gene="AR"
                /region_name="Variant"
                /note="R -> L (IN PROSTATE CANCER). /FTId=VAR_009789."
     Region     727
                /gene="AR"
                /region_name="Variant"
                /note="N -> K (IN MAIS). /FTId=VAR_009790."
     Region     728
                /gene="AR"
                /region_name="Variant"
                /note="L -> S (IN PAIS). /FTId=VAR_009791."
     Region     730
                /gene="AR"
                /region_name="Variant"
```

```
                /note="V -> M (IN PROSTATE CANCER). /FTId=VAR_004695."
     Region     732
                /gene="AR"
                /region_name="Variant"
                /note="D -> N (IN CAIS). /FTId=VAR_004696."
     Region     732
                /gene="AR"
                /region_name="Variant"
                /note="D -> Y (IN CAIS). /FTId=VAR_004697."
     Region     733
                /gene="AR"
                /region_name="Variant"
                /note="Q -> H (IN PAIS). /FTId=VAR_009792."
     Region     737
                /gene="AR"
                /region_name="Variant"
                /note="I -> T (IN PAIS). /FTId=VAR_009793."
     Region     741
                /gene="AR"
                /region_name="Variant"
                /note="W -> R (IN CAIS). /FTId=VAR_009794."
     Region     742
                /gene="AR"
                /region_name="Variant"
                /note="M -> I (IN PAIS). /FTId=VAR_004698."
     Region     742
                /gene="AR"
                /region_name="Variant"
                /note="M -> V (IN PAIS). /FTId=VAR_009795."
     Region     743
                /gene="AR"
                /region_name="Variant"
                /note="G -> E (IN CAIS). /FTId=VAR_013477."
     Region     743
                /gene="AR"
                /region_name="Variant"
                /note="G -> V (IN PAIS AND CAIS). /FTId=VAR_004699."
     Region     744
                /gene="AR"
                /region_name="Variant"
                /note="L -> F (IN CAIS AND PROSTATE CANCER).
                /FTId=VAR_009796."
     Region     745
                /gene="AR"
                /region_name="Variant"
                /note="M -> T (IN PAIS). /FTId=VAR_009797."
     Region     746
                /gene="AR"
                /region_name="Variant"
                /note="V -> M (IN PAIS). /FTId=VAR_009798."
     Region     748
                /gene="AR"
                /region_name="Variant"
                /note="A -> D (IN PAIS). /FTId=VAR_009799."
     Region     748
                /gene="AR"
                /region_name="Variant"
                /note="A -> T (IN PROSTATE CANCER). /FTId=VAR_009800."
     Region     748
                /gene="AR"
```

```
                     /region_name="Variant"
                     /note="A -> V (IN PROSTATE CANCER). /FTId=VAR_009801."
     Region          749
                     /gene="AR"
                     /region_name="Variant"
                     /note="M -> I (IN PROSTATE CANCER). /FTId=VAR_009802."
     Region          749
                     /gene="AR"
                     /region_name="Variant"
                     /note="M -> V (IN PAIS AND CAIS). /FTId=VAR_004700."
     Region          750
                     /gene="AR"
                     /region_name="Variant"
                     /note="G -> D (IN CAIS; LOSS OF ANDROGEN BINDING).
                     /FTId=VAR_004701."
     Region          750
                     /gene="AR"
                     /region_name="Variant"
                     /note="G -> S (IN PROSTATE CANCER). /FTId=VAR_009803."
     Region          751
                     /gene="AR"
                     /region_name="Variant"
                     /note="W -> R (IN CAIS). /FTId=VAR_009804."
     Region          752
                     /gene="AR"
                     /region_name="Variant"
                     /note="R -> Q (IN CAIS). /FTId=VAR_004702."
     Region          754
                     /gene="AR"
                     /region_name="Variant"
                     /note="F -> L (IN PAIS AND PROSTATE CANCER).
                     /FTId=VAR_009805."
     Region          754
                     /gene="AR"
                     /region_name="Variant"
                     /note="F -> V (IN CAIS). /FTId=VAR_004703."
     Region          755
                     /gene="AR"
                     /region_name="Variant"
                     /note="T -> A (IN PROSTATE CANCER). /FTId=VAR_009806."
     Region          756
                     /gene="AR"
                     /region_name="Variant"
                     /note="N -> S (IN PAIS). /FTId=VAR_009807."
     Region          757
                     /gene="AR"
                     /region_name="Variant"
                     /note="V -> A (IN PROSTATE CANCER). /FTId=VAR_009808."
     Region          758
                     /gene="AR"
                     /region_name="Variant"
                     /note="N -> T (PAIS; 50% REDUCTION IN TRANSACTIVATION).
                     /FTId=VAR_009809."
     Region          759
                     /gene="AR"
                     /region_name="Variant"
                     /note="S -> F (CAIS). /FTId=VAR_009810."
     Region          759
                     /gene="AR"
                     /region_name="Variant"
```

```
                /note="S -> P (IN PROSTATE CANCER). /FTId=VAR_009811."
     Region     762
                /gene="AR"
                /region_name="Variant"
                /note="L -> F (IN CAIS; LOSS OF ANDROGEN BINDING).
                /FTId=VAR_004704."
     Region     763
                /gene="AR"
                /region_name="Variant"
                /note="Y -> C (IN PAIS AND PROSTATE CANCER; PARTIAL LOSS
                OF ANDROGEN BINDING). /FTId=VAR_004705."
     Region     763
                /gene="AR"
                /region_name="Variant"
                /note="Y -> H (IN CAIS). /FTId=VAR_009812."
     Region     764
                /gene="AR"
                /region_name="Variant"
                /note="F -> L (IN CAIS). /FTId=VAR_009813."
     Region     765
                /gene="AR"
                /region_name="Variant"
                /note="A -> T (IN CAIS; LOSS OF ANDROGEN BINDING).
                /FTId=VAR_004707."
     Region     765
                /gene="AR"
                /region_name="Variant"
                /note="A -> V (IN CAIS). /FTId=VAR_009814."
     Region     766
                /gene="AR"
                /region_name="Variant"
                /note="P -> S (IN CAIS). /FTId=VAR_009815."
     Region     767
                /gene="AR"
                /region_name="Variant"
                /note="D -> E (IN CAIS). /FTId=VAR_009816."
     Region     768
                /gene="AR"
                /region_name="Variant"
                /note="L -> P (IN CAIS). /FTId=VAR_009817."
     Region     771
                /gene="AR"
                /region_name="Variant"
                /note="N -> H (IN PAIS). /FTId=VAR_009818."
     Region     772
                /gene="AR"
                /region_name="Variant"
                /note="E -> A (IN PAIS). /FTId=VAR_009819."
     Region     772
                /gene="AR"
                /region_name="Variant"
                /note="E -> G (IN PAIS). /FTId=VAR_009820."
     Region     774
                /gene="AR"
                /region_name="Variant"
                /note="R -> C (IN CAIS; LOSS OF ANDROGEN BINDING; FREQUENT
                MUTATION). /FTId=VAR_004709."
     Region     774
                /gene="AR"
                /region_name="Variant"
```

```
                /note="R -> H (IN CAIS AND PAIS; ALMOST COMPLETE LOSS OF
                ANDROGEN BINDING). /FTId=VAR_004708."
     Region     779
                /gene="AR"
                /region_name="Variant"
                /note="R -> W (IN CAIS). /FTId=VAR_004710."
     Region     780
                /gene="AR"
                /region_name="Variant"
                /note="M -> I (IN PAIS AND CAIS). /FTId=VAR_004711."
     Region     782
                /gene="AR"
                /region_name="Variant"
                /note="S -> N (IN PROSTATE CANCER; SOMATIC MUTATION).
                /FTId=VAR_009821."
     Region     784
                /gene="AR"
                /region_name="Variant"
                /note="C -> Y (IN CAIS; LOSS OF ANDROGEN BINDING AND OF
                TRANSACTIVATION). /FTId=VAR_004712."
     Region     787
                /gene="AR"
                /region_name="Variant"
                /note="M -> V (IN CAIS). /FTId=VAR_004713."
     Region     788
                /gene="AR"
                /region_name="Variant"
                /note="R -> S (IN MAIS). /FTId=VAR_009822."
     Region     790
                /gene="AR"
                /region_name="Variant"
                /note="L -> F (IN MAIS). /FTId=VAR_009823."
     Region     791
                /gene="AR"
                /region_name="Variant"
                /note="S -> P (IN PROSTATE CANCER). /FTId=VAR_009824."
     Region     793
                /gene="AR"
                /region_name="Variant"
                /note="E -> D. /FTId=VAR_009825."
     Region     794
                /gene="AR"
                /region_name="Variant"
                /note="F -> S (IN CAIS). /FTId=VAR_004714."
     Region     798
                /gene="AR"
                /region_name="Variant"
                /note="Q -> E (IN PAIS, MAIS AND PROSTATE CANCER).
                /FTId=VAR_004715."
     Region     806
                /gene="AR"
                /region_name="Variant"
                /note="C -> Y (IN PAIS). /FTId=VAR_009826."
     Region     807
                /gene="AR"
                /region_name="Variant"
                /note="M -> R (IN CAIS; LOSS OF TRANSACTIVATION).
                /FTId=VAR_004716."
     Region     807
                /gene="AR"
```

```
                /region_name="Variant"
                /note="M -> T (IN PAIS). /FTId=VAR_009827."
     Region     807
                /gene="AR"
                /region_name="Variant"
                /note="M -> V (IN CAIS; 25% ANDROGEN BINDING).
                /FTId=VAR_004717."
     Region     810
                /gene="AR"
                /region_name="Conflict"
                /note="L -> M (IN REF. 3)."
     Region     812
                /gene="AR"
                /region_name="Variant"
                /note="L -> F (IN PAIS). /FTId=VAR_009828."
     Region     814
                /gene="AR"
                /region_name="Variant"
                /note="S -> N (IN MAIS, CAIS AND PAIS). /FTId=VAR_004718."
     Region     820
                /gene="AR"
                /region_name="Variant"
                /note="G -> A (IN CAIS). /FTId=VAR_009829."
     Region     821
                /gene="AR"
                /region_name="Variant"
                /note="L -> V (IN PAIS). /FTId=VAR_009830."
     Region     827
                /gene="AR"
                /region_name="Variant"
                /note="F -> V (IN PAIS). /FTId=VAR_013478."
     Region     830
                /gene="AR"
                /region_name="Variant"
                /note="L -> P (IN PROSTATE CANCER). /FTId=VAR_009831."
     Region     831
                /gene="AR"
                /region_name="Variant"
                /note="R -> L (IN CAIS). /FTId=VAR_004719."
     Region     831
                /gene="AR"
                /region_name="Variant"
                /note="R -> Q (IN CAIS; LOSS OF ANDROGEN BINDING).
                /FTId=VAR_004720."
     Region     834
                /gene="AR"
                /region_name="Variant"
                /note="Y -> C (IN CAIS; LOSS OF ANDROGEN BINDING).
                /FTId=VAR_009832."
     Region     840
                /gene="AR"
                /region_name="Variant"
                /note="R -> C (IN PAIS). /FTId=VAR_004721."
     Region     840
                /gene="AR"
                /region_name="Variant"
                /note="R -> G (IN PAIS). /FTId=VAR_004722."
     Region     840
                /gene="AR"
                /region_name="Variant"
```

```
                /note="R -> H (IN PAIS). /FTId=VAR_004723."
     Region     840
                /gene="AR"
                /region_name="Variant"
                /note="R -> S (IN PAIS). /FTId=VAR_009229."
     Region     841
                /gene="AR"
                /region_name="Variant"
                /note="I -> S (IN PAIS). /FTId=VAR_009833."
     Region     842
                /gene="AR"
                /region_name="Variant"
                /note="I -> T (IN CAIS). /FTId=VAR_004724."
     Region     846
                /gene="AR"
                /region_name="Variant"
                /note="R -> G (IN PROSTATE CANCER). /FTId=VAR_009834."
     Region     854
                /gene="AR"
                /region_name="Variant"
                /note="R -> K (IN PAIS). /FTId=VAR_009835."
     Region     855
                /gene="AR"
                /region_name="Variant"
                /note="R -> C (IN CAIS). /FTId=VAR_004725."
     Region     855
                /gene="AR"
                /region_name="Variant"
                /note="R -> H (IN PAIS AND CAIS). /FTId=VAR_004726."
     Region     856
                /gene="AR"
                /region_name="Variant"
                /note="F -> L (IN CAIS). /FTId=VAR_009836."
     Region     863
                /gene="AR"
                /region_name="Variant"
                /note="L -> R (IN CAIS). /FTId=VAR_009837."
     Region     864
                /gene="AR"
                /region_name="Variant"
                /note="D -> G (IN CAIS). /FTId=VAR_009838."
     Region     864
                /gene="AR"
                /region_name="Variant"
                /note="D -> N (IN CAIS; LOSS OF ANDROGEN BINDING).
                /FTId=VAR_004727."
     Region     865
                /gene="AR"
                /region_name="Variant"
                /note="S -> P (IN CAIS). /FTId=VAR_009839."
     Region     866
                /gene="AR"
                /region_name="Variant"
                /note="V -> E (IN CAIS). /FTId=VAR_004728."
     Region     866
                /gene="AR"
                /region_name="Variant"
                /note="V -> L (IN PAIS). /FTId=VAR_004729."
     Region     866
                /gene="AR"
```

```
                /region_name="Variant"
                /note="V -> M (IN CAIS, PAIS AND PROSTATE CANCER).
                /FTId=VAR_004730."
     Region     869
                /gene="AR"
                /region_name="Variant"
                /note="I -> M (IN PAIS). /FTId=VAR_004731."
     Region     870
                /gene="AR"
                /region_name="Variant"
                /note="A -> G (IN PAIS). /FTId=VAR_009840."
     Region     870
                /gene="AR"
                /region_name="Variant"
                /note="A -> V (IN PAIS). /FTId=VAR_009841."
     Region     871
                /gene="AR"
                /region_name="Variant"
                /note="R -> G (IN MAIS). /FTId=VAR_009842."
     Region     874
                /gene="AR"
                /region_name="Variant"
                /note="H -> R (IN CAIS). /FTId=VAR_013479."
     Region     874
                /gene="AR"
                /region_name="Variant"
                /note="H -> Y (IN PROSTATE CANCER). /FTId=VAR_009843."
     Region     877
                /gene="AR"
                /region_name="Variant"
                /note="T -> A (IN PROSTATE CANCER; AFFECTS STEROID BINDING
                CHARACTERISTICS AND RESPONSE TO ANTIANDROGENS; FOUND IN
                BONE METASTASES). /FTId=VAR_004732."
     Region     877
                /gene="AR"
                /region_name="Variant"
                /note="T -> S (IN PROSTATE CANCER). /FTId=VAR_009844."
     Region     879
                /gene="AR"
                /region_name="Variant"
                /note="D -> Y (IN MAIS). /FTId=VAR_013480."
     Region     880
                /gene="AR"
                /region_name="Variant"
                /note="L -> Q (IN PROSTATE CANCER). /FTId=VAR_009845."
     Region     881
                /gene="AR"
                /region_name="Variant"
                /note="L -> V (IN CAIS). /FTId=VAR_009846."
     Region     886
                /gene="AR"
                /region_name="Variant"
                /note="M -> V (IN MAIS). /FTId=VAR_009847."
     Region     889
                /gene="AR"
                /region_name="Variant"
                /note="V -> M (IN CAIS AND PAIS). /FTId=VAR_009848."
     Region     890
                /gene="AR"
                /region_name="Variant"
```

```
                /note="D -> N (IN PROSTATE CANCER). /FTId=VAR_009849."
     Region     891
                /gene="AR"
                /region_name="Variant"
                /note="F -> L (IN PROSTATE CANCER). /FTId=VAR_009850."
     Region     892
                /gene="AR"
                /region_name="Variant"
                /note="P -> L (IN CAIS). /FTId=VAR_004733."
     Region     895
                /gene="AR"
                /region_name="Variant"
                /note="M -> T (IN CAIS; LOW ANDROGEN BINDING AND
                TRANSACTIVATION). /FTId=VAR_004734."
     Region     896
                /gene="AR"
                /region_name="Variant"
                /note="A -> T (IN PROSTATE CANCER). /FTId=VAR_009851."
     Region     898
                /gene="AR"
                /region_name="Variant"
                /note="I -> T (IN CAIS). /FTId=VAR_009852."
     Region     902
                /gene="AR"
                /region_name="Variant"
                /note="Q -> R (IN PROSTATE CANCER). /FTId=VAR_009853."
     Region     903
                /gene="AR"
                /region_name="Variant"
                /note="V -> M (IN PAIS). /FTId=VAR_009854."
     Region     904
                /gene="AR"
                /region_name="Variant"
                /note="P -> H (IN CAIS). /FTId=VAR_009855."
     Region     904
                /gene="AR"
                /region_name="Variant"
                /note="P -> S (IN CAIS). /FTId=VAR_009856."
     Region     907
                /gene="AR"
                /region_name="Variant"
                /note="L -> F (IN CAIS). /FTId=VAR_004735."
     Region     909
                /gene="AR"
                /region_name="Variant"
                /note="G -> E (IN PROSTATE CANCER). /FTId=VAR_009857."
     Region     909
                /gene="AR"
                /region_name="Variant"
                /note="G -> R (IN PAIS). /FTId=VAR_009858."
     Region     910
                /gene="AR"
                /region_name="Variant"
                /note="K -> R (IN PROSTATE CANCER). /FTId=VAR_009859."
     Region     911
                /gene="AR"
                /region_name="Variant"
                /note="V -> L (IN PAIS). /FTId=VAR_009860."
     Region     913
                /gene="AR"
```

```
                     /region_name="Variant"
                     /note="P -> S (IN PAIS). /FTId=VAR_004736."
     Region          916
                     /gene="AR"
                     /region_name="Variant"
                     /note="F -> L (IN CAIS). /FTId=VAR_009861."
     Region          917
                     /gene="AR"
                     /region_name="Variant"
                     /note="H -> R (IN CAIS). /FTId=VAR_009862."
     Region          919
                     /gene="AR"
                     /region_name="Variant"
                     /note="Q -> R (IN PROSTATE CANCER). /FTId=VAR_009863."
ORIGIN
        1 mevqlglgrv yprppsktyr gafqnlfqsv reviqnpgpr hpeaasaapp gasllllqqq
       61 qqqqqqqqqq qqqqqqqqet sprqqqqqqg edgspqahrr gptgylvlde eqqpsqpqsa
      121 lechpergcv pepgaavaas kglpqqlpap pdeddsaaps tlsllgptfp glsscsadlk
      181 dilseastmq llqqqqqeav segsssgrar easgaptssk dnylggtsti sdnakelcka
      241 vsvsmglgve alehlspgeq lrgdcmyapl lgvppavrpt pcaplaeckg sllddsagks
      301 tedtaeyspf kggytkgleg eslgcsgsaa agssgtlelp stlslyksga ldeaaayqsr
      361 dyynfplala gpppppppph phariklenp ldygsawaaa aaqcrygdla slhgagaagp
      421 gsgspsaaas sswhtlftae egqlygpcgg gggggggggg gggggggggg ggeagavapy
      481 gytrppqgla gqesdftapd vwypggmvsr vpypsptcvk semgpwmdsy sgpygdmrle
      541 tardhvlpid yyfppqktcl icgdeasgch ygaltcgsck vffkraaegk qkylcasrnd
      601 ctidkfrrkn cpscrlrkcy eagmtlgark lkklgnlklq eegeasstts pteettqklt
      661 vshiegyecq piflnvleai epgvvcaghd nnqpdsfaal lsslnelger qlvhvvkwak
      721 alpgfrnlhv ddqmaviqys wmglmvfamg wrsftnvnsr mlyfapdlvf neyrmhksrm
      781 ysqcvrmrhl sqefgwlqit pqeflcmkal llfsiipvdg lknqkffdel rmnyikeldr
      841 iiackrknpt scsrrfyqlt klldsvqpia relhqftfdl likshmvsvd fpemmaeiis
      901 vqvpkilsgk vkpiyfhtq
//
```


Jul 8 2003 12:22:35